3rd Neuromuscular Translational Summer School

Under auspices of EURO-NMD and TREAT-NMD

6-10 DECEMBER 2021

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	(Institut de Myologie, Group Hospitalier Pitié-Salpêtrière, Paris, France)
Silvere van der Maarel	(Leiden University Medical Center, LUMC, the Netherlands)
Andoni Urtizberea	(FILNEMUS, Filière Nationale, Neuromusculaire, 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 participants (definitely no more than 20 – or we lose the interactiveness)
- We could also target one or two industry employees (with a specific fee).

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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DECEMBER 6 (Monday)

12.00 – 12.30	**Registration and Lunch**
Session 1	**Introduction**
12.30 – 13.30	**Welcome and introduction** (T. Evangelista, S. van der Maarel, A. Urtizberea and A. Aartsma-Rus) ***Objective:*** *Introduction of participants and organizers; layout of the program and learning objectives*
13.30 – 14.30	**Overview of current state of the art of NMD management** (A. 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)*
14.30 – 15.00	**Current clinical trials in NMD** (T. Evangelista) ***Objective:*** *give participants a global overview of different types of clinical trials for NMD (15' talk, 15' discussion)*
15.00 – 15.30	**Overview of bench to bedside research** (A. Aartsma-Rus) ***Objective:*** *to outline the different steps of therapy development from idea, to proof of concept studies in model systems, to preclinical optimization studies, clinical trials, drug approval and post marketing surveillance studies (20' talk, 10' discussion)*
15.30 – 16.15	**Tea/coffee break**
16.15 – 17.00	**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 20' talks, 20' discussion)*

Session 2:	Preclinical Research
17.00 – 17.45	**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)*
17.45 – 18.00	**Introduction to TACT mock up session** (A. Aartsma-Rus)
18.00	**End of day – Q&A WITH DRINKS; MEET THE SPEAKERS**

DECEMBER 7, Tuesday

8.30 – 09.30	Self-study for TACT mock review session
09.30 -11.00	**When to move to a clinical trial? TACT mock review session** (moderated by A. Aartsma-Rus) ***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	**Coffee**
Session 3	Clinical research
11.30 – 12.30	**Introduction to clinical trials** (M. 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) *Participant 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** (Marjon Pasmooij) ***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	**Coffee break**
16.00 -17.00	**Industry perspective on drug development for rare diseases** (Thomas de Vlaam) ***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** (T. Evangelista and T.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 DRINKS; MEET THE SPEAKERS**

Faculty Dinner

DECEMBER 8, Wednesday

Session 4	Outcome measures
09.00 – 10.00	**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*
10.00- 10.30	**Showcase on outcome measure development, labelling and marketing** (Annemieke Aartsma-Rus) ***Objective:** to illustrate the consequences of using outcome measures limited to certain disease stages with regards to extrapolation and limited indications*
10:30 – 11.00	**Coffee break**
11.00 – 12.00	**Showcase on outcome measure development (PULL)** (Anna Mayhew) ***Objective**: to outline the steps and stakeholders involved in developing and validating a functional outcome measure (select one – indicate the same applies for all); 30' talk, 15' discussion*
12:00 – 13.00	**TREAT-NMD tools to facilitate clinical trials** (Michela Guglieri) ***Objective:** to gain insight in available tools and services for planning and conducting clinical trials* *Michela Guglieri ~ 45 minutes (standards of care, networking, interaction with regulators, patient registries and CTSR)* *Discussion ~15 minutes*
13.00 – 14.00	**Lunch**
14.00 – 15.00	**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)*
15.00 – 16.00	**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) 45' talk, 15' discussion)*

16.00 – 17.00	**Showcase: PROM development** (Nathalie Goemans) *Objective: explain what is involved in developing a patient reported outcome measure using the DMD PROM development as a showcase, 45' talk, 10' discussion*
17.00	**End of day – Q&A WITH DRINKS; MEET THE SPEAKERS**

DECEMBER 9, Thursday

Session 6	**Patient engagement**
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 – 11.00	**Coffee break**
11.00 – 12.30	**Translating science to the non-initiated** (Maarten Keulemans) ***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 – 15.00	**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*
15.00 – 16.30	**Presenting science to patients** ***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*

16.30 – 17.30	Presentations from each of the groups (facilitated by Silvere van der Maarel) *Objective: each of the groups presents their presentation, the other participants are the audience and listen as if they were a patient*
19.00	**Network dinner**

DECEMBER 10, Friday

09.00 – 10.30	**Participants work on preparing their final presentation**
10.30 – 12.00	**Participant presentations** (Chaired by Teresinha Evangelista and Silvere van der Maarel We ask groups of 2 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
12.00 – 12.30	Feedback and general discussion (Chaired by Annemieke Aartsma-Rus and Teresinha Evangelista)
12.30	**Lunch and departure**