3rd Neuromuscular Translational Summer School Online

Under auspices of EURO-NMD and TREAT-NMD

6-10 DECEMBER 2021

Leiden University Medical Center, the Netherlands

Program committee:

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Teresinha Evangelista	(Institut de Myologie, Groupe 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 interactivity)
- 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

3rd Neuromuscular Translational Winter School – Online 2021

DECEMBER 6, Monday

Session 1	Introduction
13.00 – 13.15	**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*
13.15 – 14.15	**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)*
14.15 – 14.30	**Tea/coffee break**
14.30 – 15.15	**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
15.15 – 16.15	**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)*
16.15 – 16.25	**Introduction to TACT mock up session** (A. Aartsma-Rus)
16.30	**End of day 1**

DECEMBER 7, Tuesday

9.00 – 10.50	**Self-study for TACT mock review session**
10.50 – 11.50	**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.*
11.50 – 12.00	**Coffee break**
Session 3	**Clinical research**
12.00 – 13.00	**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.*
13.00 – 14.00	**Lunch**
14.00 –15.00	**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*
15.00 –15.15	**Coffee break**
15.15 – 16.15	**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.)*
16.15 – 17.15	**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)*
17.15	**End of day 2**

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 – 10.15	**Coffee break**
10.15 – 11.15	**Showcase on outcome measure development (PUL)** (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*
11.15 – 12.15	**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*
12.15 – 13.00	**Lunch**
13.00 – 14.00	**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)*
14.00 – 15.00	**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)*
15.00 – 15.15	**Tea/coffee break**
15.15 – 16.15	**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*
16.15	**End of day 3**

DECEMBER 9, Thursday

Session 5	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** (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 – 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** (A. Aartsma-Rus) ***Objective:*** *A scientific paper will be presented by Annemieke and participants will listen as if they were a patient or family of a patient. Participants will be asked for constructive feedback on how to improve the presentation to make it more patient-friendly.*
16.30	**End of day 4**

DECEMBER 10, Friday

09.00 – 10.30	**Participants read eachothers blogs and give their TOP 3 selection**
	The winner will be sent a delicious bar of chocolate!
10.30	**End of Online Neuromuscular Translational Winter School**