

Skeletal muscle laminopathies – natural history and clinical trial readiness

Number	253
Date	30 OCTOBER 2020

Location: Virtual meeting

Title: 253 virtual ENMC workshop on Skeletal muscle laminopathies - natural history and clinical trial readiness

Date: 30-31 October 2020

Organisers: Dr G. Bonne (France), Dr L. Maggi (Italy), Prof. S. Quijano-Roy (France), Prof. C. Bönnemann (U.S.A)

Participants: Kate Adcock (UK), Lindsey Armstrong (Scotland), Danielle Beaumont (NL), Rabah Ben Yaou (France), Anne Bertrand (France), Sylvia Bonanno (Italy), Gisèle Bonne (France), Carsten Bönnemann (U.S.A.), Cristina Cappelletti (Italy), Robert-Yves Carlier (France), Eleonora Cugudda (Italy), Adele D'Amico (Italy), Gustavo Dziewczapolski (U.S.A.), Reghan Foley (U.S.A.), Greta Gladney, David Gómez Andrés (Spain), Marta Gomez-Garcia de la Banda (France), Uday Khire (U.S.A.), Giovanna Lattanzi (Italy), Agnieszka Madej.Pilarczyk (Poland), Lorenzo Maggi (Italy), Sebastian Maldonado (Argentina), Soledad Monges (Argentina), Andres Nacimiento Osorio (Spain), Ignacio Perez de Castro (Spain), Aty Piedra, Luisa Politano (Italy), Susana Quijano-Roy (France), Eric Schirmer (UK), Benedikt Schoser (Germany), Anneke van der Kooi (NL), Karim Wahbi (France), Howard Worman (U.S.A.)

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workshop application deadline: 1 September



253rd ENMC international workshop on Striated Muscle Laminopathies scheduled initially in March this year had to be postponed. The organizers (G Bonne, S Quijano-Roy, L Maggi, C Bönnemann) proposed to split it into two sessions, one virtual during the fall this same year and the second to be organized early next year in the usual ENMC place if the conditions allow it.

The first virtual conference was held during two afternoons on 30th and 31st October 2020. This event gathered 30 participants from Europe and America, including healthcare providers and researchers from Argentina, France, Italy, Netherlands, Spain UK and USA, as well as patients' and representatives from four advocacy associations (Cure-CMD, Fundacion Marcio Contra las laminopatías (FAM), Muscular Dystrophy UK (MD-UK), Associazione Italiana Distrofia Muscolare di Emery Dreifuss (AIMED)).

The meeting was opened by G Bonne (Paris, France), who welcome participants and gave an overview of the meeting main objectives: to increase knowledge in these diseases, to improve their diagnosis, clinical management and follow-up, to facilitate the identification of biomarkers and to move forward in possible therapeutic strategies.

The first afternoon session was dedicated to parallel working group (WG) meetings performed in several virtual rooms: 1) The Basic science & outcome measures WG chaired by G Bonne (Paris) was focused on patho-mechanisms of the striated (i.e., skeletal and cardiac) muscle laminopathies, eventual therapeutic targets including CRISPR and potential biochemical and imaging biomarkers; 2) The Heart WG (K Wahbi, Paris, France) presented recently published studies in adult striated muscle laminopathies and available data in pediatric patients; 3) The Clinical pediatric WG (S Quijano-Roy, Garches, France, and C Bönnemann, Bethesda, USA) allowed to update cohorts of patients followed by the different participating teams and to present the results of a recent worldwide international retrospective study with clinical and genetic data from more than 150 pediatric patients; and 4) The clinical adulthood WG (L Maggi, Milan, Italy) presented retrospective data of the adults affected with striated muscle laminopathies and discussed the most frequent measurements and outcomes identified in several studies. In all clinical WGs patients, families and

and people with neuromuscular disorders

March 2020 -

09:38



ENMC

White Paper

February 2020 -

08:00



The 2nd

report of the special ENMC workshop on the role of the neuromuscular

patient in

Shared-Decision-

Making!

10 June

2019 - 19:33



ENMC

bibliometric

report

published

1 April

2019 - 12:30



Publication

on the impact of

25 years ENMC

workshops

12

March 2019 -

09:16



Press

release – ENMC

publication about

the role of

patients in NMD

research

7

February 2019 -

18:00



group representatives had a time to express their ideas, needs and expectations. After these brainstorming sessions, all WG reconvened in a single virtual room and WG leaders presented the most important data and results, what allowed to state at best a state of the art.

The second afternoon session was important to structure and discuss about all presented data. Innovative therapies seem now to be closer and clinical readiness is becoming of great importance. Clinical and complementary motor, respiratory and cardiac outcome measures were listed by different teams to reach a consensus; potential biomarkers including biochemical and imaging techniques were described. The results of the retrospective pediatric worldwide study and other data in adult laminopathies provided a useful starting point to design prospective clinical studies in view of future therapeutic trials.

The meeting ended with a contribution of the patients' group representatives, where they and their relatives could share their experiences in daily life and highlighted their difficulties, unmet needs and expectations.

Although it was the first time that this ENMC workshop was run online, participants found it a successful experience which allowed experts in clinical and basic research, and patients and associative representatives to review updated data, interact in real-time and identify priorities and face future perspectives.

This virtual meeting has been an excellent opportunity to update and deepen our knowledge of natural history of laminopathies, promoting the international research and collaborative studies to better characterize and follow our patients, while we wait to be able to meet again in the face-to-face meeting in 2021.

The workshop organisers are preparing a survey to be circulated before the next planned virtual session in February 2021.

April 19th 2021:

The 253rd ENMC workshop on Laminopathies continued on 19 April 2021 via 4 online break-out rooms: Phenotyping, Biomarkers, SOC motor and contractures, and Trial Readiness. The groups will continue their work and will come back together (virtually!) in two months' time. Thanks to all participants for the interesting discussions and great progress made!



New ENMC

Research

Director 121

January 2019 -

15:26



Announcement:
As of 2018, ENMC
relocated its
workshop
venue23 July
2018 - 17:16

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