253rd virtual ENMC International Workshop: Striated Muscle Laminopathies

Due to the COVID-19 SARS2 worldwide pandemic context, the 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 three advocacy associations (Cure-CMD, Fundacion Marcio Contra las laminopatias (FAM), 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 advocacy group representatives had a time to express their difficulties, 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.