RevEal the burdeN on daily life for myotonic dyStrophy patients due to myotoniA: preliminary results of the ENSA survey

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It is well accepted in the myotonic dystrophy (DM) community that myotonia can place a substantial burden on daily life for people with DM1 or DM2. However, the negative impact of DM is poorly quantified, and there is little patient-reported information about the contribution of myotonia to the daily burden of DM. ENSA is a patient-reported, international, online survey conducted in 2023 to investigate the impact of myotonia on daily life in people with DM.

Adults (≥18 years) with DM1 or DM2 in Europe, the UK or North America complete the ENSA anonymised survey, publicized via an outreach campaign. ENSA includes 32 questions on self-reported descriptions of DM symptom onset, time to medical consultation, myotonia frequency/location, muscle weakness, fatigue, daytime sleepiness, gastrointestinal/cardiorespiratory symptoms, disease management, treatment, and impact of DM on daily life.

Preliminary analyses of data collected until April 12, 2023, revealed that 169 DM patients (or their representatives) in 10 countries had completed the ENSA survey to date. The mean (range) age of respondents was 45.6 (11–77) years; n=89 (53%] were female; n=157 (93%) had DM1. Myotonia was reported by 156 (92%) patients: 130 (77%) currently had myotonia and 26 (15%) had experienced myotonia in the past; 13 (8%) had never experienced myotonia. Of those with myotonia currently, 67/130 (52%) were not receiving any drug treatment for this symptom. Updated data will be presented. In summary, here we present methodology and preliminary data from ENSA. The overall findings of ENSA dataset will aim to help quantify the impact of myotonia on DM1 and DM2 patients' daily lives. They also aim to increase awareness and understanding of the ongoing need for effective management of this debilitating symptom.

Disclosures:

Zozulya-Weidenfeller is employed by Lupin. Other authors received honoraria from Lupin as consultants during the ENSA creation.