Poster presentation at the 7th International Myotonic Dystrophy Conference - September 2009

Diagnostic odyssey of myotonic dystrophy type 2 (DM2) patients

Background

Delays in receiving an accurate diagnosis can have a significant impact on the burden of disease for patients, on family planning, and on the management of their symptoms. Colleagues from Finland have hypothesized that many DM2 patients are misdiagnosed because symptoms of DM2 often overlap with those of other more common disorders.

We investigated diagnostic delay in myotonic dystrophy type 1 (DM1) and DM2 patients enrolled in the National Registry by analyzing patient-reported data and reviewing patient medical records. We analyzed data related to patients' onset of symptoms and diagnosis of DM. This included information about patients' first symptoms, initial misdiagnoses, and diagnostic exams.

Results

There was an average delay in the diagnosis of DM2 of 15 years. This delay was twice the length of the delay for the diagnosis of DM1. Data from our retrospective chart review indicate that 22% of genetically confirmed DM2 patients received a misdiagnosis prior to obtaining the correct diagnosis of DM2. These inaccurate diagnoses included inclusion body myositis, limb girdle muscular dystrophy, and rheumatoid arthritis. DM2 patients also received significantly more diagnostic exams, such as EMGs, muscle biopsies, and DNA testing compared to DM1 patients. Most DM2 patients reported their first symptom as leg weakness (29.3%), whereas the most common symptom was grip myotonia in DM1 (37.5%).

Conclusion

Diagnostic delay was significantly longer in DM2 compared to DM1. The impact of this delay is evidenced by the increased number of diagnostic exams and frequent misdiagnoses received by DM2 patients. Additional studies are necessary to help better understand the potential broad spectrum of presenting symptoms in DM2 to facilitate more timely recognition and diagnosis of DM2. Opportunities also exist to use the resources of the Registry to further study the impact of diagnostic delay in DM on pain, economic and psychosocial burdens of disease, and symptom management.