INTRODUCTION

• Myotonic Dystrophy Type 1 (DM1) is a hereditary, autosomal dominant disorder. It is a rare, slowly progressive, and disabling multi-systemic disease.

• DM1 is the most common form of adult muscular dystrophy, affecting 8.2 in 100,000 worldwide, and leads to myotonia (slowed relaxation of contracted skeletal muscle), muscle weakness and wasting, as well as gastrointestinal, central nervous system, and cardiac abnormalities.

• For a complex disease like DM1, patient-reported outcome (PRO) measure could serve as a useful tool to examine disease burden and to assess treatment effect.

OBJECTIVES

- To identify the PRO measures used to capture disease burden and quality of life in patients with DM1.
- To identify how disease burden and quality of life changes over time in patients with DM1, using longitudinal studies.

METHODS

- Literature search: Comprehensive literature reviews and evaluation were conducted for relevant publications in English. Search database includes: PubMed, MEDLINE, clinicaltrial.gov, and clinicaltrialsregister.eu.


- Keywords for clinical trial databases: [Myotonic dystrophy]

- PRO measures were then validated through the PROQOLID database.

- Independent screening was conducted by two reviewers, and any discrepancies between reviewers were reconciled by further discussion to reach an agreement.

- The search was conducted on July 19th, 2014

RESULTS

- Overall, 2527 DM1 patients were enrolled in the articles and clinical trial databases reviewed.

- The PROs used were mostly general and as such only targeted sequelae of the disease. Sequelae targeted include pain, muscle weakness, mobility/disability, sleep, fatigue, quality of life, etc. An exception to this piecemeal approach is the MDHI instrument, which captured several domains of the disease.

- MDHI is a disease-specific measure designed to estimate overall disease burden and impact of key symptomatic themes in DM1 patients. However, more studies are needed to evaluate the validation and responsiveness of this instrument.

- Frequently-used generic measures were the Epworth Sleepiness Scale and the SF-36, applied in seven (33%) studies. Even though these scales measure different domains, study findings show deterioration across these domains in DM1 patients.

- Further, only three studies were longitudinal and thus were able to capture changes in quality of life and other burden over time in DM1 patients. Findings from these longitudinal studies indicate significant deterioration in the quality of life in DM1 patients over time, marked with decreased ambulation and increased dependence on others.

- Disease-modifying therapies are needed to reduce disease progression and improve quality of life in patients with DM1.

REFERENCES

