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Characterization of pain in myotonic dystrophy type 1

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Abstract

Background

Myotonic dystrophy type 1 or DM1 is an autosomal dominant, multisystem muscular dystrophy with an incidence of 1 per 8000, caused by pathological noncoding CTG repeat expansion in the DMPK gene (1).

Pain is considered a common symptom in all types of neuromuscular disorders (NMD) with a prevalence of 30–90%, present both in adults and children (2,3). However, this patient's complaintis often overlooked in DM1.

The aim of the study was to evaluate the intensity and interference with daily living activities (ADL) of pain in patients with DM1.

Methods

We recruited patients with a genetic diagnosis of DM1, assessed by our evaluation protocol, including collection of anamnestic and anthropometric data, and pain assessment tool as Brief Pain Inventory (BPI) with its two indexes: Severity Index (BPI-SI) and Interference Index (BPI-II). Based on SI and II, our population was divided in a mild group and a moderate-severe group, using a cut-off value <4 for both outcome measures. We evaluated between-group differences for the following outcome measures: Tinetti Performance Oriented Mobility Assessment (POMA); Functional Independence Measure (FIM); Fatigue Severity Scale (FSS).

Results

We evaluated 45 patients, 23 males and 22 females, with mean age of 47.33 +17.15 and mean BMI of 26.84 +4.84. According to pain measures, 19 patients reported mild pain (42.22%) and 17 moderate to severe pain (37.77%), 17 patients (37.77%) minimal pain interference in the ADL, 19 moderate to severe interference (42.22%). The average scores obtained in the outcomes were: 19.88 +7.02 at the POMA, 112.82 +11.69 at the FIM and 37.24 +19.12 at the FSS. Based on the severity index, a statistically significant difference was found between the two groups for the fatigue outcome, while for the interference index, all outcomes showed a statistically significant difference among groups.

Conclusion

Our results demonstrated that pain significantly affects balance and gait performance, fatigue and functional independence in patients with DM1.

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