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ScienceDirect

Neuromuscular Disorders 25 (2015) 141–148

www.elsevier.com/locate/nmd

Muscle force, balance and falls in muscular impaired individuals with myotonic dystrophy type 1: A five-year prospective cohort study

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Received 24 June 2014; received in revised form 4 November 2014; accepted 10 November 2014

Abstract

Individuals with myotonic dystrophy type 1 (DM1) have progressive muscle weakness with gait and balance impairments. We explored prospectively the natural history of muscle force, gait, balance, balance confidence and walking ability in muscular affected individuals with DM1. After five years data from 43 individuals (m/f:18/25) were analysed. All measures of balance showed statistically significant deterioration ($p < 0.001$) with averaged yearly loss of function by 3–4%. In the group as a whole, loss of muscle force was statistically significant in all lower limb muscles measured after five years: changes relative to baseline force were median −6% to −18%. For males muscle force loss was statistically significant in all leg muscles, but only in hip flexors for women. After five years 100% of the men had fallen during the previous year and 67% three times or more, in contrast only 60% of the women had fallen in the previous year and 36% three times or more. The proportion of individuals seeking medical care the previous year, after falling, was more than doubled after five years, albeit the number of falls had not changed. Awareness of this increased risk of falls is important for caregivers and patients.

Keywords: Myotonic dystrophy; Observation; Self-reported falls; Postural balance; Muscle strength; Physiotherapy

1. Introduction

Myotonic dystrophy type 1 (DM1) is an inherited neuromuscular disorder with the highest prevalence among adults, 9–18/100.000 inhabitants [1,2]. Functional impairments in multiple organ systems are common, including cognitive deficits and fatigue [3,4]. Myotonia and muscle weakness in the distal muscles of the leg and arm are the main characteristics of the muscle affection in individuals with DM1 [3].

In individuals with DM1 an increased risk of stumbles and falls has been shown [5]. The main factor for this is suggested to be the slowly progressive weakness in the leg muscles [5]. The cognitive deficits and the fatigue [6] could contribute to the impaired postural control resulting in frequent stumbles and falls. There is a need for a closer knowledge of the evolvement of the balance impairment and falls of the individual with DM1.

Only a few studies of the progression of muscle weakening and impairments in individuals with DM1 have been conducted [7–11]. By using manual muscle testing an average decline of 0.95% per year in muscle strength has been shown in a cross-sectional study of 50 individuals with DM1 [7]. The study evaluated muscle strength loss per year of disease duration. The decline was more rapid in the distal muscles with no significant difference between genders (0.99% in females and 1.54% in men per year). The same study evaluated muscle force with quantitative measurements using dynamometry in hand grip, lateral pinch, elbow extensors, hip flexors and ankle dorsiflexors, showing a decline of 1% (elbow extensors in females) to 3% (hand grip in men) per year of disease duration. Furthermore, a gender effect noted was that women were weaker than men in all muscles [7]. Hand grip, lateral pinch and neck muscle force have been evaluated in a longitudinal study over two years with dynamometry and strain gauge, showing very small but significant force increases, that according to the authors probably are due to normal variations or a training effect [8]. All other muscles were evaluated with manual muscle testing, through full range, and showed a strength decline [8]. Another longitudinal study, with 158 consecutive individuals with DM1, showed a decline in handgrip force of 1.18 kg/year in women and 1.61 kg/year in men [9]. A ten year prospective evaluation of 75 patients with non-congenital and

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http://dx.doi.org/10.1016/j.nmd.2014.11.004
0960-8966/

17 patients with congenital myotonic dystrophy showed a generalised weakness in both types, but progressive in the non-congenital type, only [10]. When performing a selenium–vitamin E study on patients with myotonic dystrophy, the authors concluded that a useful set of measurements to follow functional deterioration in this disorder would be hand-grip strength and maximal walking speed [11]. To our knowledge no prospective study on performance-based measures of balance in connection with leg muscle force using handheld dynamometry has been performed in individuals with DM1.

A prior mapping study including balance confidence, performance-based measures of balance, leg muscle force, walking and number of falls confirmed the increased risk of falling [12]. One factor of importance for increased number of falls was shown to be muscle weakness. A still remaining question is the natural course of the physical impairments. This is important for future studies on intervention effects in individuals with DM1.

The objectives of this study were to explore, in muscular impaired individuals with DM1, the natural history in muscle force, gait and performance-based measures of balance, as well as patient reported balance confidence, falls and walking ability.

2. Patients and methods

2.1. Patients

All 72 eligible individuals with genetically proven DM1 at the Neuromuscular Centre, Sahlgrenska University Hospital, were invited by letter and/or by phone call to participate in a cross-sectional study [12]. All fifty-one patients who participated in this cross-sectional study were available for the present longitudinal study. In the previous study the inclusion criteria were: genetically confirmed non-congenital DM1 diagnosis; age between 20 and 60 years; and ability to perform Timed Up & Go (TUG) [13]. The exclusion criterion was other disorder that could affect postural control. Exclusion criteria for the present study were: (1) MIRS grade 1 or 2 (see section 2.3) or (2) inability to perform TUG in less than 30 seconds at baseline. The reason for the former criterion was that patients with MIRS 1 and 2 have the mild form of DM1 and thus have very little muscle symptoms. The mild form of DM1 is also reasonably stable over time, in our experience. The reason for the TUG criterion was that this cut-off in elderly individuals distinguishes between individuals dependent or independent in basic transfers [13]; and that patients walking so slowly would be likely to be a wheelchair user in a three year perspective, and need other forms of support. These narrowed criteria were due to the interest in postural control evaluation of weak but still independently ambulant patients, at risk for falls. Data from the mapping study were used as baseline data in the present study. The patients gave their written informed consent to participate in the study and knew that they could refuse further participation without any impact on further treatment. The Regional Ethical Review Board in Gothenburg, Sweden approved the study (Dnr 248-06 and Dnr 601-11).

2.2. Procedures

An experienced physiotherapist (EH), although having met some of the patients in the clinic during the five years, examined all patients at baseline and after five years, blinded to previous results. The self-assessments and the performance-based measures of balance were performed in a standardised order; first the self-assessments followed by the physical examinations. The examiner demonstrated the positions in the balance tests. Resting pauses were allowed and encouraged. The timing was performed with an electronic stopwatch with an accuracy of 1/100 second.

2.3. Measurements and assessments

An assessment and classification with Muscular Impairment Rating Scale (MIRS) was performed [14]. The **MIRS** is an ordinal scale with five grades, with the definitions of the grades as follows: 1 – no muscular impairment; 2 – minimal signs, as ptosis and nasality, no distal weakness except digit flexor weakness; 3 – distal weakness, no proximal weakness except in elbow extensors; 4 – mild to moderate proximal weakness; and 5 – severe proximal weakness. For a more detailed definition see the original paper [14].

The **isometric muscle force** was assessed by the maximal amount of resistance in an isometric muscle effort with a handheld gauge meter (Mecmesin Basic Force Gauge 1000N, Chauvin Arnaux Group), and was recorded in newtons (N). The "break" method according to Phillips et al. [15] and Bäckman et al. [16] was used. The assessed muscle groups in this study were the hip flexors, knee extensors, knee flexors and the ankle dorsiflexors. The hip flexors and ankle dorsiflexors were assessed in supine position, according to Phillips et al. [15], and the calf was supported with a pad during the ankle dorsiflexor assessment to get a free calcaneus. The knee extensors were measured in the sitting position, and the knee flexors were measured in the prone position, according to Bäckman et al. [16]. The test–retest reliability of the handheld dynamometry in patients has been reported as Pearson's r 0.96–0.99 for the same muscle groups as was measured in the present study [17] and ICC 0.91–0.97 in another study in individuals with spinal muscular atrophy [18].

Timed 10-m walk (10mMAX): Walking at maximum speed was measured in a long corridor with an even surface over 10 meters with a still-standing start and a "flying" finish to a target 2.5 meters beyond the mark at 10 m [19–22]. The stopwatch was started on the word "Go" ("Ready-Steady-Go"). The patients were instructed to take their shoes and callipers off to minimise the risk of shoe difference bias at follow-up. The patients could choose to walk barefoot or in socks. An exception was made to allow AFO use when this was the only possible way a patient could walk 10 meters. Handheld walking aids were allowed when needed. All patients were well acquainted with the test. The test has shown test–retest stability in individuals with DM1 (ICC 0.94) [23].

Timed Up & Go (TUG): The patient rose with arm support from a seated position in a chair of normal height (44–45 cm) with arms, walked at a comfortable and safe pace to a mark on