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Trunk muscle involvement is most critical for the loss of balance control in patients with facioscapulohumeral muscular dystrophy

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ABSTRACT

Background: Although it is known that muscle weakness is a major cause of postural instability and leads to an increased incidence of falls in patients with neuromuscular disease, the relative contribution of lower extremity and trunk muscle weakness to postural instability has not been studied well.
Methods: We determined the relationship between muscle fatty infiltration and sagittal-plane balance in ten patients with facioscapulohumeral muscular dystrophy. Sagittal-plane platform translations were imposed in forward and backward directions on patients with facioscapulohumeral muscular dystrophy and healthy controls. Stepping thresholds were determined and kinematic responses and center-of-mass displacements were assessed using 3 dimensional motion analysis. In the patients, magnetic resonance imaging was used to determine the amount of fatty infiltration of trunk and lower extremity muscles.
Findings: Stepping thresholds in both directions were decreased in patients compared to controls. In patients, significant correlations were found for fatty infiltration of ventral muscles with backward stepping threshold and for fatty infiltration of dorsal muscles with forward stepping threshold. Fatty infiltration of the rectus abdominis and the back extensors explained the largest part of the variance in backward and forward stepping thresholds, respectively. Center-of-mass displacements were dependent on intensity and direction of perturbation. Kinematic analysis revealed predominant ankle strategies, except in patients with lumbar hyperlordosis.
Interpretation: These findings indicate that trunk muscle involvement is most critical for loss of sagittal-plane postural balance in patients with facioscapulohumeral muscular dystrophy. This insight may help to develop rehabilitation strategies to prevent these patients from falling.

1. Introduction

Facioscapulohumeral muscular dystrophy (FSHD) is a common hereditary myopathy that causes weakness of the face, shoulder girdle, trunk and leg muscles due to progressive fatty infiltration (Padberg, 1982; Padberg & van Engelen, 2009; Patijn, 1983). Although nearly all muscles of the trunk and lower extremities can be affected, individual muscle involvement varies greatly between patients (Lunt & Harper, 1991; Padberg, 1982). Generally, disease progression is slow, but as soon as a muscle becomes affected the process towards total muscle fattening occurs relatively fast (Janssen et al., 2014). The degree of fatty infiltration shows a high correlation with both muscle strength and the Ricci (Ricci et al., 1999) clinical severity score (Iosa et al., 2007; Janssen et al., 2014; Rijken et al., 2014). As a consequence, patients with FSHD suffer from postural instability and show a high incidence of falls, mainly in the forward and backward directions (Horlings et al., 2009a; Pieterse et al., 2006). Since FSHD does not cause disturbances of muscle tone, motor coordination, sensibility, or loss of joint range of motion, it constitutes a good model to examine the influence of specific patterns of muscle weakness on the control of posture and balance (Horlings et al., 2008, 2009b).

In a previous study from our group, Horlings and coworkers have included patients with distal spinal muscular atrophy (SMA; predominantly distal weakness of arms and legs) and patients with limb–girdle muscular dystrophy (LGMD; predominantly proximal weakness of trunk, arms and legs) as models to investigate the contribution of proximal versus distal leg muscle weakness to postural instability. By contrasting responses to balance perturbations, it was demonstrated that participants with distal weakness had larger sagittal-plane center-of-mass (CoM) displacements compared to those with proximal weakness. Hence, loss of distal leg muscle strength seemed to be a critical factor in sagittal-plane instability (Horlings et al., 2009b). However, in this study, the contribution of trunk muscle weakness to postural instability was not addressed, although trunk muscles

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play a critical role in the recovery from balance perturbations (Carpenter et al., 2008; Henry et al., 1998; Horak & Nashner, 1986; Preuss & Popovic, 2010; Runge et al., 1999).

In persons with FSHD, it is well known that abdominal muscles are afflicted relatively early in the course of the disease, whereas back extensor muscles were not believed to show major involvement (Padberg et al., 1991; Patijn, 1983). In a recent study, however, it was demonstrated that the back extensor muscles did exhibit substantial fatty infiltration in many FSHD patients (85%) as well, including people in early disease stages (Rijken et al., 2014). We expect that this frequent involvement of the back extensors, in addition to the abdominal muscles, will greatly affect core stability with, consequently, profound impact on postural control. Similarly, a previous study also suggested a possible association between trunk muscle weakness in people with FSHD and decreased control of upper body movement during walking (Iosa et al., 2010).

Only in healthy subjects, the effects of lumbar extensor versus ankle plantar flexor weakness on balance recovery following a forward postural perturbation have been contrasted by inducing localized muscle fatigue by repetitive movement. The maximum perturbation that could be withstood without stepping was not significantly affected by localized muscle fatigue in either muscle group, but CoM excursions increased in both conditions (Davidson et al., 2009). Although it is unknown whether fatigue-induced muscle weakness is similar to FSHD-related loss of strength, we assume that the weakness of ~30% as induced in the paper of Davidson et al., was only minor compared to the loss of muscle strength that patients with FSHD experience (e.g. almost 50% lower quadriceps strength in ambulatory FSHD patients than in controls (Bachasson et al., 2014)). Hence, the specific effects of (severe) leg versus trunk muscle weakness on postural stability have not been thoroughly investigated.

The aim of this study was to determine whether the affliction of trunk or lower limb muscles is ultimately most critical for the loss of postural control by studying the responses to sagittal-plane balance perturbations in patients with FSHD. We hypothesized that trunk muscle involvement would be more critical than lower extremity muscle involvement for sagittal-plane balance control. To this aim we performed radiological assessments of muscle fat infiltration (FI) in ten patients with FSHD of different disease severities as well as balance assessments with full-body kinematic recordings of these FSHD patients and ten healthy control subjects.

2. Methods

2.1. Participants

Ten adults with genetically confirmed FSHD, based on blood DNA testing, were recruited via the rehabilitation and neurology departments of our university hospital during a period of six months. We aimed to include a range of disease severities; from patients who did not yet experience postural problems during activities of daily life on the one end of the spectrum to severely affected patients who were barely able to walk at the other end. Specific exclusion criteria were: the presence of other neurological diseases affecting muscle strength, balance or sensibility; contra-indications for physical exercise; metal implants (contra-indication for MRI); and pregnancy. Ten healthy controls of similar age and gender were also included. This study was approved by the local medical–ethical committee. All subjects gave written informed consent.

2.2. Procedure

During an intake visit a physiatrist checked the inclusion and exclusion criteria and determined the clinical severity score (CSS) as described by Ricci et al. (1999) for each patient. To grade balance performance in the patients, the Berg Balance Scale (BSS) was used. Following the intake visit, the radiological assessments were performed on the same day. The balance assessments in the movement laboratory were done within 8 weeks after the intake. Healthy controls did not undergo radiological assessments and were only tested in the movement laboratory.

2.3. Radiological assessments

The patients were examined with a 3.0 Tesla (T) MR system (Skyra; Siemens, Erlangen, Germany) using the body coil. Imaging of the trunk and lower extremity was performed using transverse and coronal turbo spin echo T1-weighted sequences. Transverse images were obtained from four different regions; sequences of ten slices each of the trunk at the level of vertebra L4, the pelvis between the symphysis and the anterior superior iliac spine, the upper leg at 15 cm proximal to the apex of the patella, and the lower leg at 15 cm below the patella (TR 750, TE 9.4, turbo factor 3, matrix 256 × 256, slice thickness 5 mm, slice gap 1 mm). In these regions, also coronal imaging was performed from the anterior to posterior side of the body (TR 650, TE 10, turbo factor 2, matrix 512 × 512, slice thickness 5 mm, slice gap 0.5 mm). The degree of FI was scored by one experienced musculoskeletal radiologist (JR) on a modified four point scale for 70 muscles (35 on each body side) within the regions covered by MRI. This semi-quantitative score correlates inversely to muscle strength (Schwartz et al., 1988). The FI of the muscles was graded as: 1. normal appearance, with a homogeneous low signal intensity of the muscle; 2. early 'moth-eaten' appearance (i.e. scattered areas or beginning confluence of high signal intensity consistent with fat; less than fifty percent of muscle affected); 3. scattered areas of high signal intensity as well as confluent zones of hyperintensity; more than fifty percent of muscle affected; and 4. 'washed-out' appearance consistent with complete replacement of the muscle by high signal intensity fat and connective tissue, with a rim of fascia remaining (end-stage disease). The presence of muscle atrophy was not considered in the semi-quantitative scoring system, given the large variation in muscle size even in healthy subjects. Because of the minor differences between left and right, for each muscle the FI score was calculated as the average of the MRI FI scores of the left and right body side. For each patient, the number of muscles with an average FI score ≥ 2 was determined.

A lumbar hyperlordosis is often seen in patients with FSHD (Lee et al., 2009; Tawil & Van Der Maarel, 2006a) and may affect the kinematic response of the body to a perturbation. Therefore, weight-bearing total spine conventional radiographs were made in both the anterior–posterior and lateral direction to assess the spinal curvature. The lumbar lordosis was measured in the sagittal plane as the angle between the inferior endplate of vertebra L5 and the superior endplate of vertebra L1 (Cobb's angle) (Harrison et al., 2001).

2.4. Balance assessments

To quantify the maximum sustainable balance perturbation we used an instrumented movable platform to perform standardized balance perturbations (Radboud Falls Simulator). Subjects were exposed to anterior and posterior translations with an acceleration period of 300 ms, a constant-speed period of 500 ms, and a deceleration period of 300 ms. An anterior translation causes a backward perturbation and a posterior translation a forward perturbation of the body. Prior to the perturbations, subjects held their hands on their waist. Subjects wore a safety harness attached with a cable to a sliding rail at the ceiling that allowed frictionless movement. They were instructed to try their hardest to overcome the perturbations while keeping their feet in place. A trial was considered successful if the subject recovered balance without stepping or grabbing the surrounding handrails.

One first trial in both directions was performed to familiarize subjects with the type of perturbations. Thereafter, perturbations were administered according to a defined trial sequence, which existed of backward and forward perturbations in an unpredictable order. Three trials were included for each level of intensity, starting at an intensity of 0.25 m/s^2,