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# Mobility assessment of patients with facioscapulohumeral dystrophy

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**Abstract**

*Background*. Facioscapulohumeral muscular dystrophy is the third most common form of inherited myopathies with a prevalence of 1:20,000. Since both muscle involvement and disease progression are heterogeneous and unpredictable, quantitative assessment tools are needed to evaluate the effects of pharmacological and physical training treatments.

*Methods*. The instrumented movement analysis of 12 patients with facioscapulohumeral dystrophy and 12 control subjects was conducted using a 9-camera stereophotogrammetric system and 2 force platforms. Subjects performed four tasks of different difficulties: arm movement, level walking, step ascending, and squatting. Manual muscle test, clinical severity scale and magnetic resonance imaging were used to clinically assess the patients.

*Findings*. Walking speed and centre of mass vertical displacement during squatting were reduced in patients and can be used to assess their motor capacity. Features common in the patient sample were: the reduction of shoulder range of motion, the excessive ankle plantar-flexion during walking and step ascending, and the reduction of knee flexion–extension moment during squatting. These parameters were correlated with magnetic resonance imaging results at relevant structure level and can be used to assess the corresponding body functioning. Furthermore, instrumented movement analysis was able to distinguish from normal controls also a group of patients in which clinical assessments did not show any obvious abnormalities and had been evaluated as normal.

*Interpretation*. The quantitative assessment tool devised in this study provides suitable information in terms of both motor capacity and impairment severity of patients with facioscapulohumeral dystrophy, and, thus, encouraging its use for the evaluation of therapeutic trial outcomes for this disease.


*Keywords:* Facioscapulohumeral muscular dystrophy; Movement analysis; Motor capacity; Impairment severity; Shoulder range of motion; Gait; Step ascent; Squat

## 1. Introduction

Facioscapulohumeral muscular dystrophy (FSHD) is an autosomal dominant progressive myopathy causing motor impairment and mobility limitation (Padberg, 1982). Its estimated prevalence of 1:20,000 makes it the third most common form of inherited myopathies after Duchenne and myotonic muscular dystrophies (Emery, 1991). Typical characteristics are asymmetric muscle weakness with early involvement of facial and scapular muscles and eventual spreading to upper limb, abdominal, pelvic, and lower limb muscles. Onset age and muscle involvement pattern and severity, characterized in most patients by the coexistence of affected and apparently unaffected muscles, is highly variable (Lunt and Harper, 1991; Padberg, 1982). Although the disease genetic defect has been localized to the long arm of chromosome 4 (region 4q35) (Upadhyaya et al., 1992; Wijmenga et al., 1992) the pathophysiological mechanisms responsible for the progressive muscle impairment are still unknown (Tawil and Van Der Maarel, 2006).

Treatment procedures for patients with FSHD mainly rely on providing functional loss compensations. Few

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pharmacological trials have been performed (Rose and Tawil, 2004), but their efficacy has not been proven. Recent observations suggest that physical training could improve motor performance, at least in the short term, but more robust evidence is needed to prove this hypothesis (Olsen et al., 2005; van der Kooi et al., 2004). A major limitation in designing clinical protocols useful for therapeutic trials in FSHD is due to patient heterogeneity and to the inability to define the disease natural history. Semiquantitative and qualitative scales based on the manual muscle test (MMT) and motor function measurements have been proposed for the assessment of the disease effects (Berard et al., 2005; Kilmer et al., 1995; Ricci et al., 1999). These scales, however, lack sensitivity and objectivity and are affected by floor/ceiling effects (Eagle, 2002; van der Kooi et al., 2005). Recently, the magnetic resonance imaging (MRI) was proposed as a clinical assessment tool for FSHD patients (Olsen et al., 2006). However, MRI provided essential information only at body structure level in terms of changes of muscle fibre structure, such as muscle replacement and edemas, and the impairment was quantified by image visual inspection.

Hence, an objective evaluation of subject's functioning changes due to FSHD is still lacking in the clinical community. To this purpose, instrumented movement analysis has been proposed as a tool for an objective evaluation in relation to different muscular disorders (Armand et al., 2005; Reynolds et al., 1999; Wright et al., 1995). This approach has been associated with motor tasks more complex than level walking, such as obstacle avoidance or step ascent and descent, for sensitivity enhancement (Pavan et al., 2005). Nevertheless, to the authors' knowledge, instrumented movement analysis of FSHD has been limited so far to the description of the kinematics of gait and upper limb tasks (Moreno Izco et al., 2005).

The objective of this study was to devise a movement analysis protocol specific to patients with FSHD, able to provide an objective assessment in spite of the above mentioned heterogeneity of the disease progression and the presence of compensation strategies. This protocol was expected to provide parameters for the assessment of both motor activity (assessment at whole body level) and body function (assessment at body structure level) in terms of motor capacity and impairment severity, respectively (WHO, 2001). Both upper and lower limb functions were tested and movement data were collected while subjects executed motor tasks of different difficulty. Relevant results were compared with standard clinical parameters to verify whether an added value associated with the use of the instrumented approach was observed.

## 2. Methods

### 2.1. Subjects

Twelve patients affected by FSHD (patient group, PG: $n = 8$ males, age range = 26–58 years, body mass index = 27 (SD = 6) kg/m$^2$) for whom the clinical diagnosis had been confirmed by the molecular genetic test of double EcoRI-BlnI digestion (Deidda et al., 1996) (EcoRI size range = 16–35 kb) and twelve healthy subjects (control group, CG: $n = 7$ males, age range = 23–47 years, body mass index = 24 (SD = 3) kg/m$^2$) were involved in this study after informed consent was given.

### 2.2. Clinical assessment

Disease duration, defined as the number of years from symptoms onset, was recorded for each patient and was used as indicator of disease progression. Even though this parameter is subjective, it can be considered valid in this study because of the uniformity of the group since it lacks cases with disease onset both at an advanced age and at infancy (before 12 years of age). Only one patient (P1 in Table 1) was still unaware of the disease symptoms at 48 years of age.

Lower limb joint flexion and extension were evaluated by the manual muscle test (MMT) and a score was assigned according to the Medical Research Council Scale (MRC, 1976) (MMT-score, ranging from 0 = no movement, no visible or palpable contraction to 5 = segment movement through full range of motion (RoM) against gravity and ability to hold against resistance). Average MMT-scores were computed at muscular group level, at joint level (average of relevant muscular group level MMT-scores), and at whole body level ($MMT_t$-score, average among all joint level MMT-scores).

A 10-grade clinical severity scale (Ricci et al., 1999) was adopted to assign a score to the overall level of mobility limitations (clinical-score ranges from 0.5 = facial weakness to 5 = wheelchair bounded). According to this scale, clinical-score $\leqslant 2$ was assigned to patients with facial and shoulder muscles weakness and higher scores (>2) were assigned to patients showing also pelvic and lower limb muscles weakness. The subgroup of patients with clinical-score $\leqslant 2$ will be indicated as $PG_1$ and the remaining subgroup as $PG_2$ in what follows.

Table 1
Clinical parameters for the patient group

| Patient | Clinical-score | Disease duration | $MMT_t$ | $MRI_t$ | Age (y.o.) |
|---|---|---|---|---|---|
| P1 | 1 | 0 | 5.0 | 0.0 | 48 |
| P2 | 1 | 4 | 5.0 | 0.0 | 30 |
| P3 | 1 | 12 | 5.0 | 0.0 | 26 |
| P4 | 1.5 | 5 | 5.0 | 0.2 | 45 |
| P5 | 1.5 | 7 | 5.0 | 0.1 | 26 |
| P6 | 3 | 19 | 5.0 | 0.8 | 36 |
| P7 | 3 | 16 | 4.6 | 0.6 | 34 |
| P8 | 3 | 18 | 4.6 | 1.1 | 37 |
| P9 | 3 | 28 | 4.7 | 0.8 | 44 |
| P10 | 3.5 | 6 | 4.3 | 1.3 | 44 |
| P11 | 3.5 | 22 | 3.8 | 0.9 | 57 |
| P12 | 3.5 | 37 | 4.7 | 0.4 | 58 |

Patients P1–P5 constitute group $PG_1$.