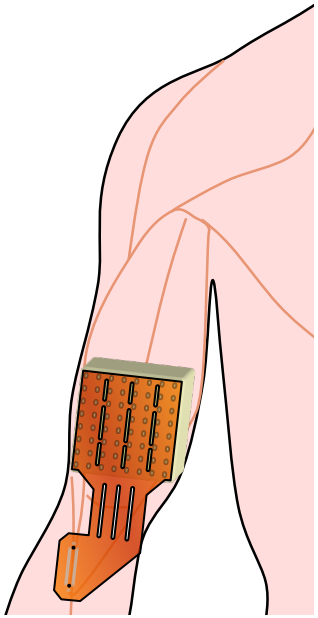
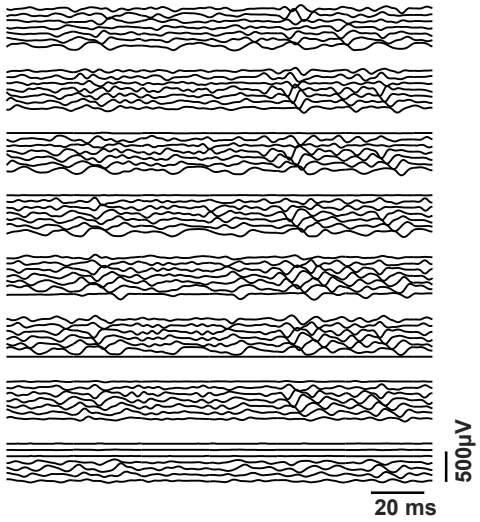
Scuola universitaria professionale
della Svizzera italiana
SUPSI

UNIVERSITÀ
DI PAVIA

Increased resistance towards fatigability in patients with facioscapulohumeral muscular dystrophy (FSHD)



Giornata della
Ricerca in
Medicina Umana



Matteo Beretta-Piccoli, PhD

Rehabilitation Research Laboratory 2rLab and Criams-Sport Medicine Centre Voghera

Foreword

This cross-sectional study was conducted in compliance with the *standards of Good Clinical Practice* (International Council for Harmonization - ICH-E6), and with the approval of the ethics committee of the University of Pisa.

I would like to express my gratitude
to all the FSHD patients and their families for participating in the study.

I would like to acknowledge all the authors:
Luca Calanni · Massimo Negro · Giulia Ricci · Cinzia Bettio · Marco Barbero · Angela Berardinelli · Gabriele Siciliano · Rossella Tupler · Emiliano Soldini · Corrado Cescon · Giuseppe D'Antona

This study was published in the Eur J Appl Physiol 2021
https://doi.org/10.1007/s00421-021-04650-3

1. BACKGROUND

- FSHD is characterized by a distinctive pattern of skeletal muscle weakness, affecting i.a. facial, shoulder and upper arm muscles (Mul et al., 2016; Tawil, 2018; Ricci et al., 2016).

- Progression of the disease is associated with atrophy and fatty infiltration of the muscle tissue (Mul et al. 2017).

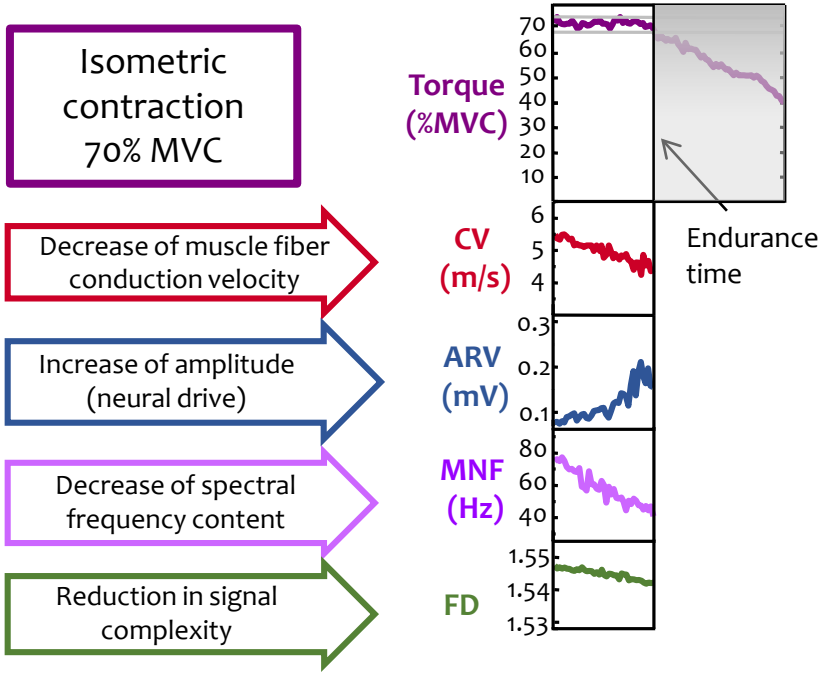
- One of the symptoms of FSHD with the highest prevalence (93.8%) is *fatigue* (Hamel et al., 2019), although its occurrence has been poorly investigated.

- According to Kluger et al. (2013) fatigue is defined as a symptom characterized by feelings of tiredness and weakness, in which physical and cognitive functions are impaired, because *of interactions between performance and perceived fatigability*.

1. BACKGROUND AND AIM

- *Performance fatigability* refers to the decline in an objective measure of performance (Enoka and Duchateau, 2016).

- It can be *indirectly* assessed using parameters associated to the surface electromyographic (sEMG) signal (Rampichini et al., 2020).



- The objective of this study was to investigate whether patients fatigue differently than healthy controls (HC) during sustained isometric elbow flexions.

We hypothesized that since FSHD is outlined by a transition from fast-glycolytic to slow-oxidative muscle fibres (Celegato et al. 2006), patients would show less performance fatigability compared to controls.

2. METHODS – participants

- Data collection began in 2013 and was completed in 2019.
- 17 patients with FSHD (8 women) and 17 healthy controls were recruited.
- Inclusion criteria were age of ≥ 16 years, a clinical or genetic diagnosis of FSHD, and enrollment in the Italian National Registry for FSHD.
- Exclusion criteria were wheelchair bound at selection, use of corticosteroids, severe cardiac and respiratory dysfunction, and psychological/psychiatric disorders.
- A diagnosis of FSHD had to be confirmed by DNA testing (Lemmers et al. 2012) at the University of Modena and Reggio Emilia (Italy).

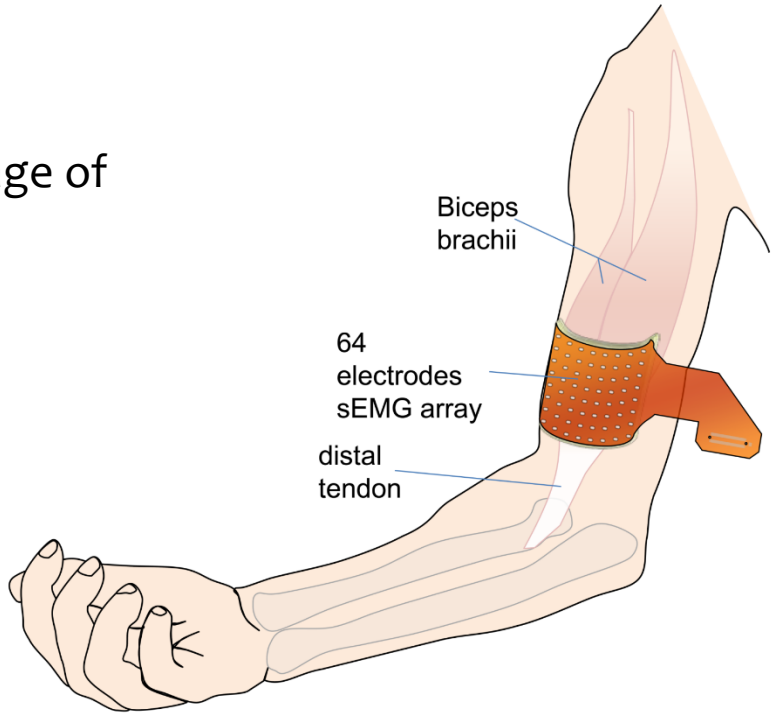
2. METHODS – fatiguing task and sEMG

- Participants performed two maximal voluntary isometric contractions (MVC) of the dominant biceps brachii, followed by:

1) a 20% MVC for 2 min, and
2) a 60% MVC held until exhaustion.

- Muscle weakness was characterized as a percentage of predicted values.

- Normalized slopes (wrt their initial values) of:
  - ARV (central factors)
  - MNF (central and peripheral factors)
  - CV (peripheral factors)
  - FD (central factors)

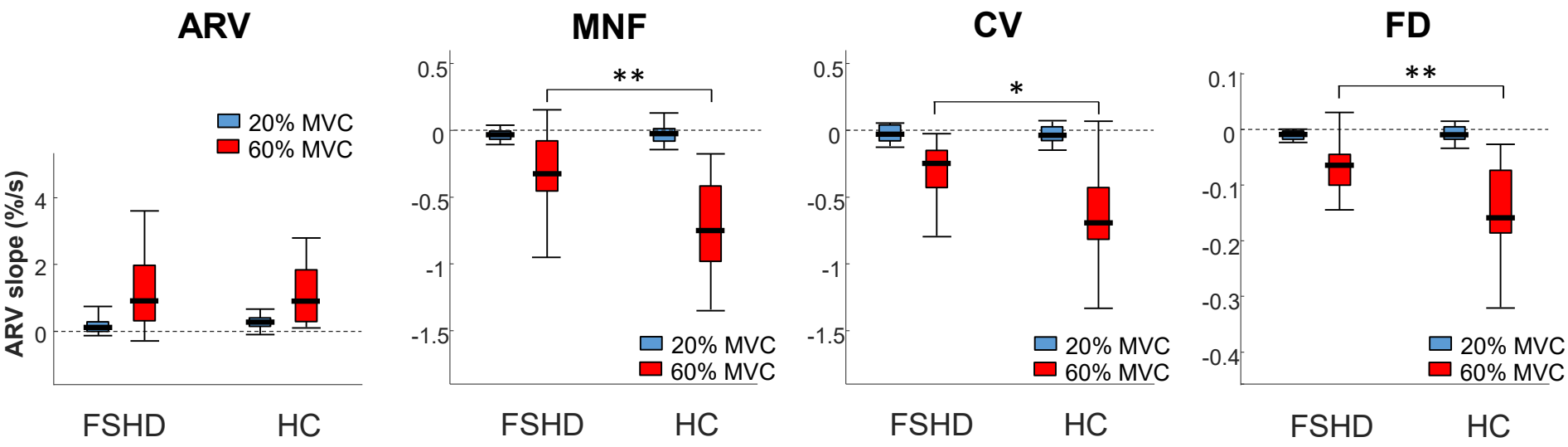


and compared between the two groups, using the Mann-Whitney U test.

3. RESULTS

Neural drive (ARV) was not different among the two groups.

The negative slopes of MNF, CV and FD showed a less steep decline at 60% MVC in the FSHD group.



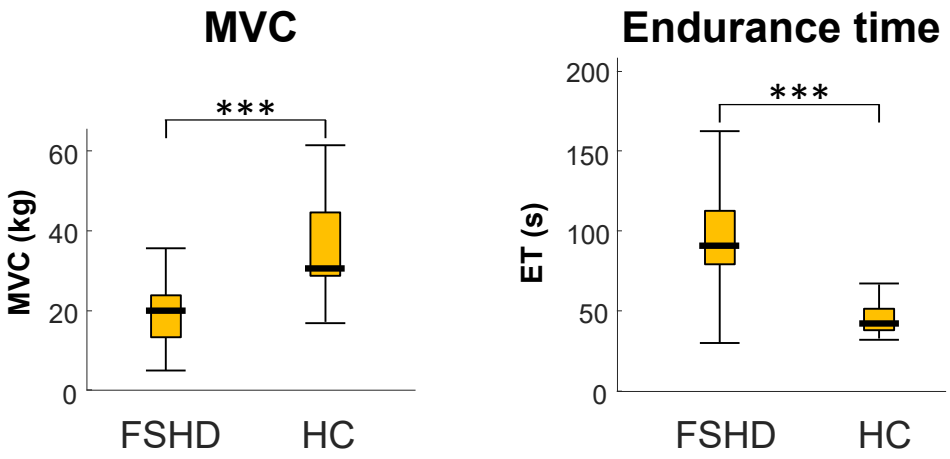
* p<0.05, ** p<0.01, *** p<0.005

Effect size analysis revealed high values for MNF slope and FD slope during 60% MVC contraction (r estimates 0.56 and 0.51, respectively).

3. RESULTS

The exerted maximal strength (MVC) was lower in the FSHD group.

The endurance time was longer compared to the HC group.



*** p<0.005

In patients with FSHD, the maximal strength was 31.3% significantly lower than the predicted value (19.8 kg vs 28.8 kg; p<0.01).

Effect size analysis revealed high values for MVC and endurance time (*r* = 0.62 and 0.63, respectively).

4. CONCLUSIONS

I. Compared to the controls, in FSHD patients, the fatigability parameters changed similarly, but with a lower decrease in the slopes, during sustained isometric elbow flexions, suggesting a reduction in performance fatigability.

II. FSHD patients exerted less force with respect to the controls, which may be caused by the muscle weakness.

III. Patients seem to be less prone to get fatigued (higher endurance time), probably due to a strong alteration of the sarcomeric contractile properties, preferentially of fast-glycolytic muscle fibers.

Thank you for your attention!