

A research paper on the characteristics and impact of pain on quality of life (QoL) in patients with facioscapulohumeral dystrophy (FSHD) was recently published in *Muscle & Nerve* (2017). In the paper, analysis is carried out on data collected via the UK FSHD Patient Registry.

The aim of this paper is to determine the frequency, localisation and intensity of pain in the FSHD1 population and to evaluate the influence of pain, age, sex, disease duration and ambulatory status on quality of life (QoL). Recent studies have indicated that pain may be present in the majority of FSHD patients and that pain negatively impacts on quality of life (QoL) and adds an increased disease burden. At present the data available is scarce, often not FSHD specific and is usually clinician reported.

The published paper is called "Chronic Pain Has a Strong Impact on Quality of Life In Facioscapulohumeral Muscular Dystrophy" and was written by Dr Germán Morís and Dr Teresinha Evangelista (Newcastle University, UK) with help from fellow researchers based across the UK. In total 398 UK FSHD Patient Registry participants were included in the analysis, a summary of which can be viewed below:

## Key Features of the Study

- Questionnaires were patient reported without interference of clinicians.
- The Patient Registry, uses Patient Reported Outcome Measures (PROMS) to collect data on pain and quality of life (QoL).

## Tools Used for Measurement

### Short Form of the McGill Pain Questionnaire (SF-MPQ)

Consists of 15 descriptors (11 sensory and 4 affective) which are rated on an intensity scale as; 0 = none, 1 = mild, 2 = moderate or 3 = severe.

### Universal Pain Assessment Tool (UPAT)

Developed by the multi-disciplinary team at Newcastle University was also used. UPAT is in the process of validation and it includes sections on pain intensity, current pain and chronic pain; in addition, to questions regarding medication and other non-pharmacological therapies

### Individualised Neuromuscular Quality of Life Questionnaire (INQoL)

Patients completed INQoL, a widely used and well-validated neuromuscular disease specific measure of QoL

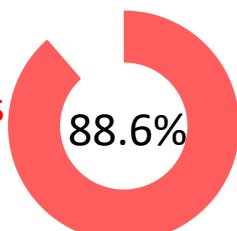
## Key Findings - SF-MPQ (383 participants)



- SF-MPQ sensory pain score was 5.0 (9.0), the median affective pain score was 1.0 (3.0), and total pain score was 6.0 (12.0).
- The sensory score in females (6.0) was significantly higher ( $p < 0.05$ ) than in males (3.0).
- There was a tendency for people with onset of symptoms at an earlier age, or longer disease duration to report more intense pain on SF-MPQ.

## Key Findings - UPAT (367 participants)

325 (88.6 %) participants reported experiencing current pain.



- The majority of patients reported that pain was localised at shoulders and lower back in 263 (72.3 %) and 259 (71.5 %) patients respectively.

- Severe lower back pain was reported more frequently in females (33 patients, 17.7 %) than in males (14 patients, 11.2 %) with statistical significance ( $p < 0.05$ ).
- Of the 365 people providing answers, 203 (55.6 %) reported experiencing chronic pain, 69 (30.4 %) reported this pain as severe.

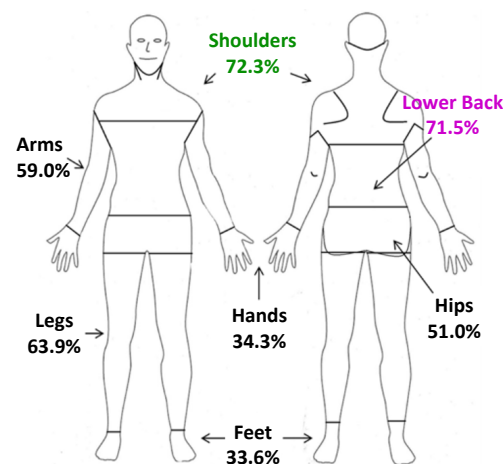


Figure 1: Description of the localisation and the severity of the current pain in each localisation expressed in percentage.

## Key Findings - INQoL (340 participants)

- The median overall INQoL score was 53.1 (34.3) implying relatively moderate sickness-related dysfunction in this group of FSHD1 patients.
- The sub-domains with the highest median scores, indicating the greatest impact on QoL were muscle weakness 63.2 (26.4), body image 59.7 (51.4) and activities 57.4 (58.6).
- The least impact on QoL is in the areas of muscle locking 10.5 (22.4) and relationships 20.4 (16.7).
- The weakness score in males 73.7 (36.8) was significantly higher than in females 68.4 (47.4) ( $p < 0.01$ ).

## Summary of Findings

- The UK FSHD Patient Registry has shown that it is able to collect a large amount of data over a short period of time.
- This study demonstrates that pain is highly prevalent in people with FSHD1.
- Female FSHD1 patients experience pain more frequently than males.
- No correlation was seen between chronic pain and current age, age of onset or disease duration.
- Chronic pain and the severity of it were not significantly correlated with the D4Z4 fragment size or motor function.
- More than 90 % of the patients reported taking medication.
- INQoL domains having the biggest impact on quality of life (QoL) were muscle weakness, activities and body image.
- Patients perceive a deterioration in QoL with the progression of the disease; so that, the younger the age of onset and the longer the disease duration, the higher the patients' perception of disability.