



Manifestations and Impact of Facioscapulohumeral Muscular Dystrophy (FSHD): Preliminary Results from a Survey of FSHD Patients

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Background

- FSHD is one of the most common muscular dystrophies.¹
- FSHD affects roughly 1 in 8-20,000 people amounting to approximately 20,000 individuals in the US and 450,000 individuals worldwide.²
- Muscles affected include those of the face, shoulders, arms, and lower extremities.³
- Limited studies detail patient-reported impact of muscle weakness on functional activities.

Methods

- This anonymous survey was developed with input from clinical experts, the FSH Society, and patient focus groups.
- The 42 questions were designed to assess patient characteristics (15), disease impact (16), and clinical trial participation (11).
- Questions were included regarding upper and lower extremity strength and function that might inform clinical trial outcome measure selection.
- The survey was sent to the FSH Society's patient contact database of 2,000 with 440 completed surveys received, including 388 patients and 52 caregivers/friends.
- Results from 388 patient responses collected from 11 Dec 2016 to 18 Feb 2017 are presented here.

Patient Characteristics and Population

- Patients were mostly female (53%) and mostly from the US (86%).
- Mean disease duration from diagnosis was 18.5 years (range 0-58) (Table 1).
- Although 75% of patients had been genetically tested for FSHD, 57% of all patients did not know which type they had.
- Most patients (67%) use some form of assistive device for ambulation such as orthotics, walkers and powered mobility.

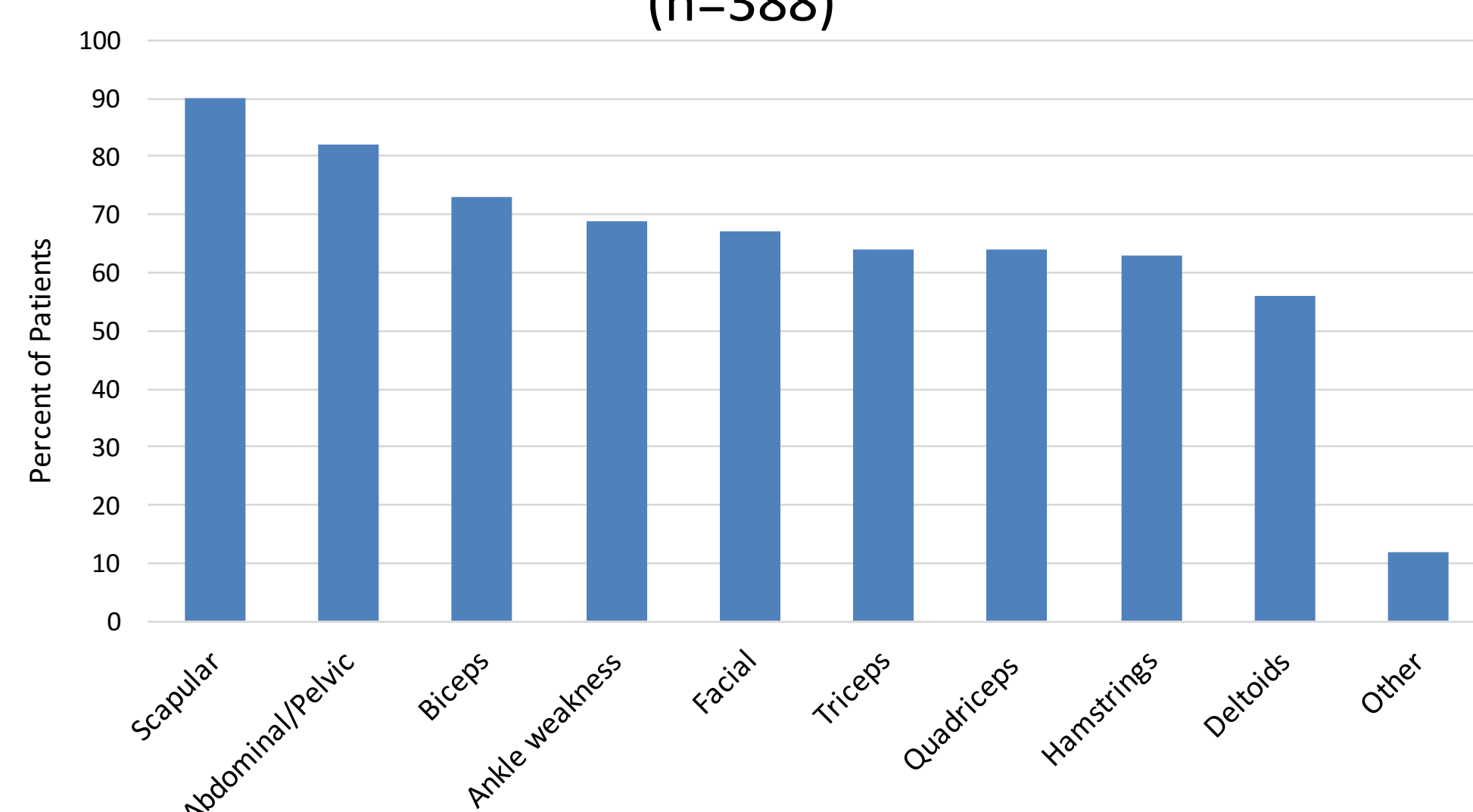
Table 1: Age of Onset and Diagnosis (n=388)

Patients	Mean	Range	n
Age of symptom onset	22	1 – 70	279
Age of diagnosis	33	1 – 75	322
Age at present	52	15 – 86	375

Disease Signs and Symptoms

- The majority of patients reported muscle weakness commonly associated with FSHD including 67% with facial, 90% with scapular and 73% with biceps weakness (Fig. 1).
- Other frequently reported muscle weaknesses included abdominal/pelvic weakness in 82% and ankle weakness/foot drop in 69% of patients.
- Biceps weakness was bilateral in 71% of patients and foot drop was bilateral in 43%.

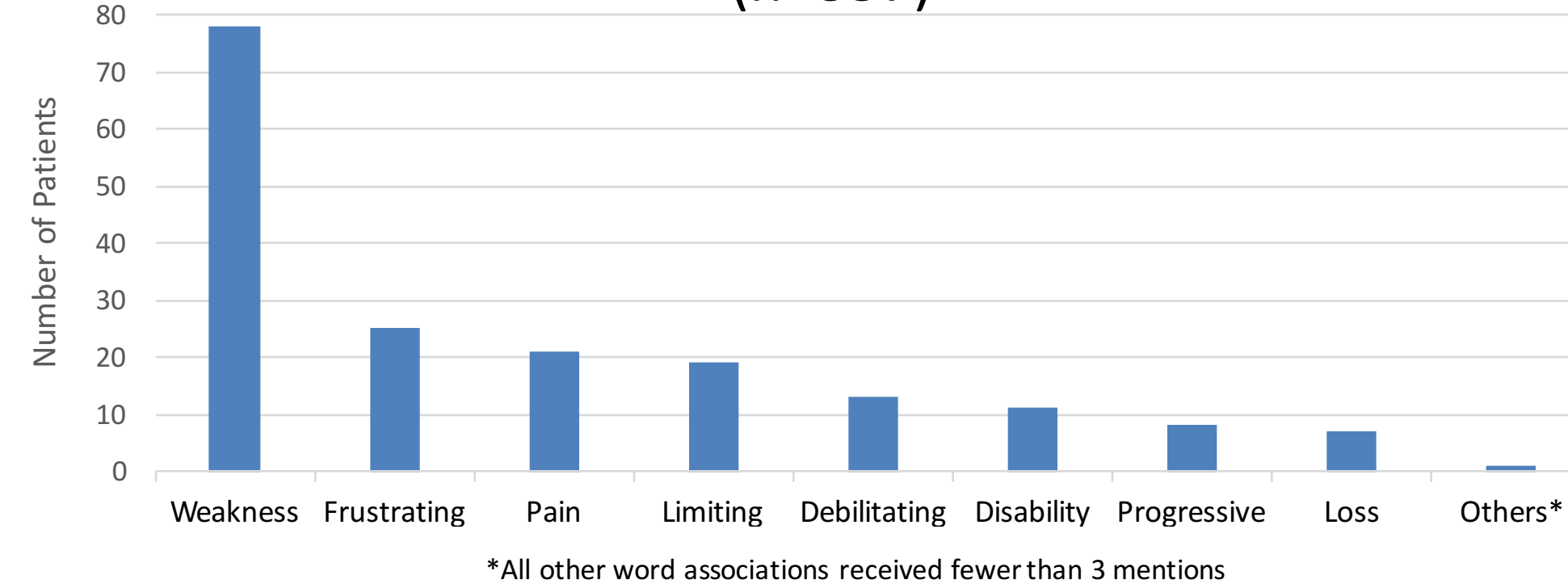
Figure 1: Muscle Weakness Manifestations due to FSHD (n=388)



Disease Challenges and Quality of Life

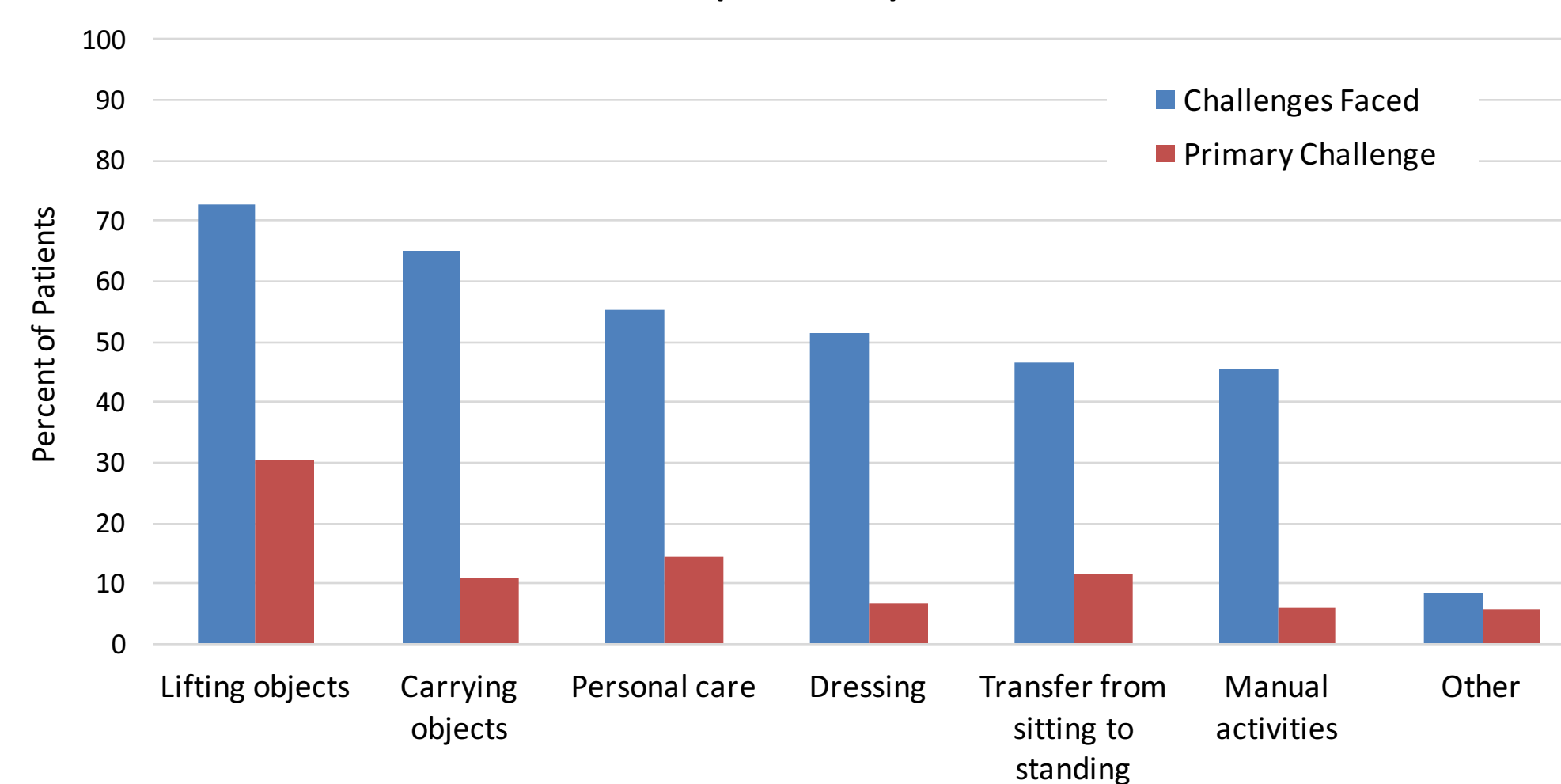
- "Weakness" was most often cited as the one word patients associate with FSHD (Fig. 2).

Figure 2: FSHD Word Association (n=357)



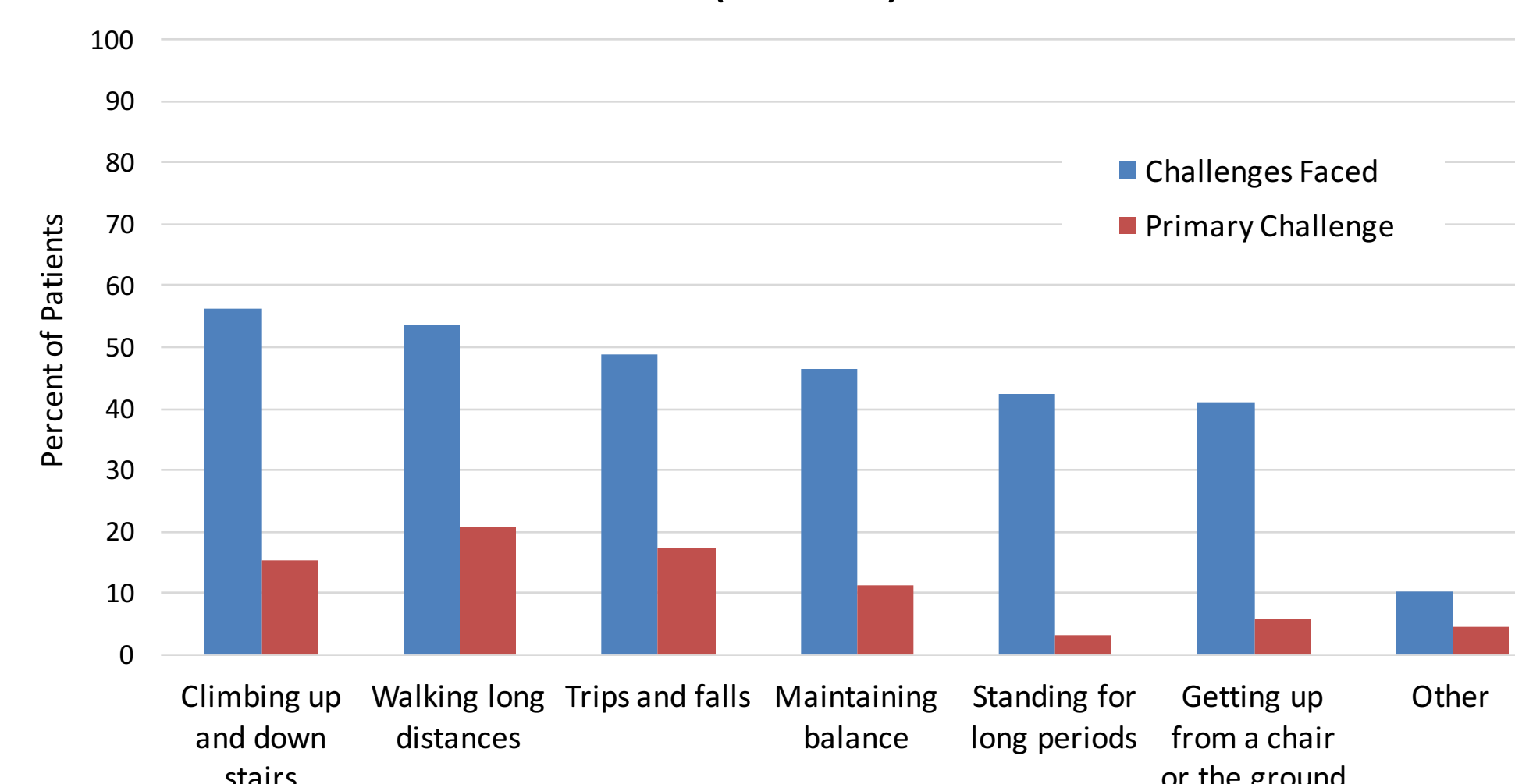
- The most common challenges associated with biceps weakness were lifting objects (73%), carrying objects (65%) and personal care (55%) (Fig. 3).
- Patients most reported lifting objects as the primary challenge of biceps weakness (31%).

Figure 3: Challenges due to Biceps/Upper Arm Weakness (n=388)



- The most common challenges associated with foot drop were climbing stairs (56%), walking long distances (53%) and trips and falls (49%) (Fig. 4).
- Patients most reported walking long distances as the primary challenge of foot drop (21%).
- The median and mean number of near falls was 3 and 13 (range 0-606), while actual falls to the ground was 0.12 and 1.1 (range 0-20) per month.
- Primary factors contributing to patient falls were loss of balance/coordination (65%), weakness in other muscles (59%), and foot drop (51%).

Figure 4: Challenges due to Foot Drop/Ankle Weakness (n=388)



- Patients' quality of life was affected "Moderately" to "Very much" by weakness in arm/shoulder (88%), foot/leg (80%), and core/abdominal muscles (81%).
- Fatigue and pain also frequently impacted patients' quality of life (Table 2).

Table 2: Factors Affecting Patient Quality of Life (n=388)

	Very much	Moderately	Slightly	Not at all
Foot/leg weakness	58%	22%	14%	7%
Arm/shoulder weakness	53%	35%	11%	1%
Core/abdominal weakness	51%	30%	13%	6%
Fatigue	35%	39%	21%	4%
Pain	19%	34%	33%	15%
People's lack of understanding	19%	25%	33%	24%
Having to keep my FSHD secret	10%	8%	16%	67%
Loss of facial expression	9%	20%	35%	38%
Breathing issues	6%	16%	28%	50%
Hearing loss	5%	11%	25%	59%
Speech impairment	2%	6%	28%	64%

Clinical Operations and Healthcare Utilization

- The top motivations for patients to participate in clinical trials included helping others and the feeling that they are doing all they can to help.
- Other motivating factors included the idea of advancing science and being the first to try a treatment (Table 3).

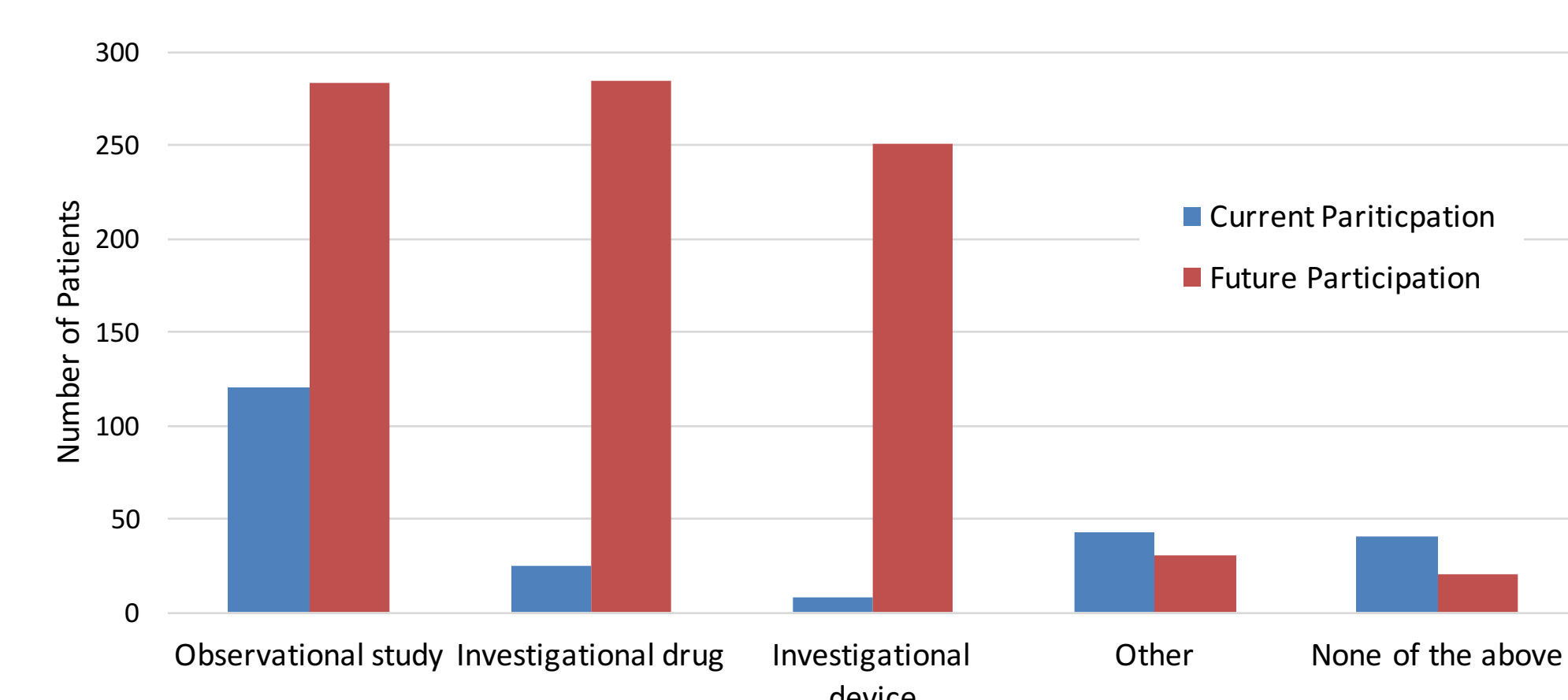
Table 3: Motivators for Clinical Trial Participation (n=388)

Even if it might not help me, it will help others some day	69%
The feeling that I'm doing everything I can do to help	57%
The idea of advancing science	57%
The chance to be first in line to try a treatment	42%
I'll only try something that's proven, so I wouldn't volunteer	5%
Other	4%
I'm not seeking a drug to cure FSHD, so I wouldn't volunteer	2%

Note: Patients could select more than one motivator for clinical trial participation.

- An observational study was the most common type of clinical trial in which patients had participated in previously (31%) (Fig. 5).
- Patients were most interested in future clinical trials involving an investigational drug (73%), an observational study (73%) or an investigational device (65%).

Figure 5: Current and Anticipated Future Clinical Trial Participation (n=388)



Note: Patients could select more than one type of clinical study.

Conclusions

- Patients' survey responses suggest a high prevalence of upper and lower body muscle weakness.
- Key muscle weakness challenges included avoiding falls and being able to lift and carry objects.
- Pain and fatigue are additional challenges possibly related to muscle weakness.
- Therapies aimed at improving foot drop or weakness in the biceps may be important to patient daily functioning and quality of life.
- Many patients demonstrated interest in participating in various clinical trials.

References and Acknowledgements

References

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3. Wang L, Tawil R. Current Neurology and Neuroscience Reports 2016;16(7).

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