A Qualitative Study to Understand the Duchenne Muscular Dystrophy Experience from the Caregiver/Patient Perspective

Victoria Brown[1], Elizabeth Merikle[1], Kelly Johnston[1], Ivana Audhya[2], Katherine Gooch[2], Linda Lowes[3]

[1]Labcorp Drug Development, Gaithersburg, Maryland, USA; [2]Sarepta Therapeutics, Inc., Cambridge, MA, USA; [3]Nationwide Children's Hospital, Columbus, OH, USA

BACKGROUND

- Duchenne Muscular Dystrophy (DMD) is a rare condition that occurs in boys and is characterized by deterioration of muscle, resulting in loss of ambulation, decreased upper limb mobility, and impaired cardiorespiratory function. No cure for DMD exists; treatment with corticosteroids primarily focuses on management of symptoms and complications.
- Understanding the experience of living with DMD from the patient perspective is necessary to improve disease management, provide better therapeutic options, and to help inform development and selection of patient-reported outcome measures for use as endpoints in DMD studies.
- To date, only two qualitative studies have identified and described symptoms and functional issues that matter most to patients with DMD[1,2]. Both studies included a small sample of ambulatory boys (< 10 patients and/or caregivers) and none have explored how symptoms may vary across different stages of ambulation.

OBJECTIVE

- To generate qualitative evidence on caregiver and patient experiences with symptoms of DMD and their impacts on overall function and daily life in ambulatory and non-ambulatory patients.
- Information was also gathered on caregiver and patient expectations of future treatments for DMD and anticipated benefits.

METHODS

- 46 dyads (caregiver and DMD patients aged 4 to 22 years) in the United States participated in 60-minute semi-structured video interviews.
- Participants were recruited from patients attending the muscular dystrophy or neuromuscular disorders clinics at Nationwide Children's Hospital. Further outreach was conducted via email invitation through the Parent Project Muscular Dystrophy patient advocacy registry.
- To be eligible, patients needed to be male and ≥4 years old or older with a genetically-confirmed DMD diagnosis. Caregivers needed to be ≥18 years old and the primary caregiver of a patient.
- Interviews were conducted via videoconference by two qualitative researchers using a semi-structured interview guide that included concept elicitation to characterize the caregiver and patient perspectives of their DMD experience, its impact on overall function and daily life, which symptoms pose the biggest challenges, and expectations for new treatments.
- Interview transcripts were analyzed using thematic analysis. Differences in the patient experience by ambulation status were examined. Findings from the thematic analysis informed development of a conceptual model of DMD.

RESULTS

- 28 ambulatory boys (mean age=8.7 years, SD=3.35) and 18 non-ambulatory boys (mean age=11.3 years, SD=3.27) participated in the study.
- The majority of caregivers were mothers of individuals with DMD. 4 caregivers had one other child with DMD. Other characteristics of the caregivers are provided in **Table 1**.

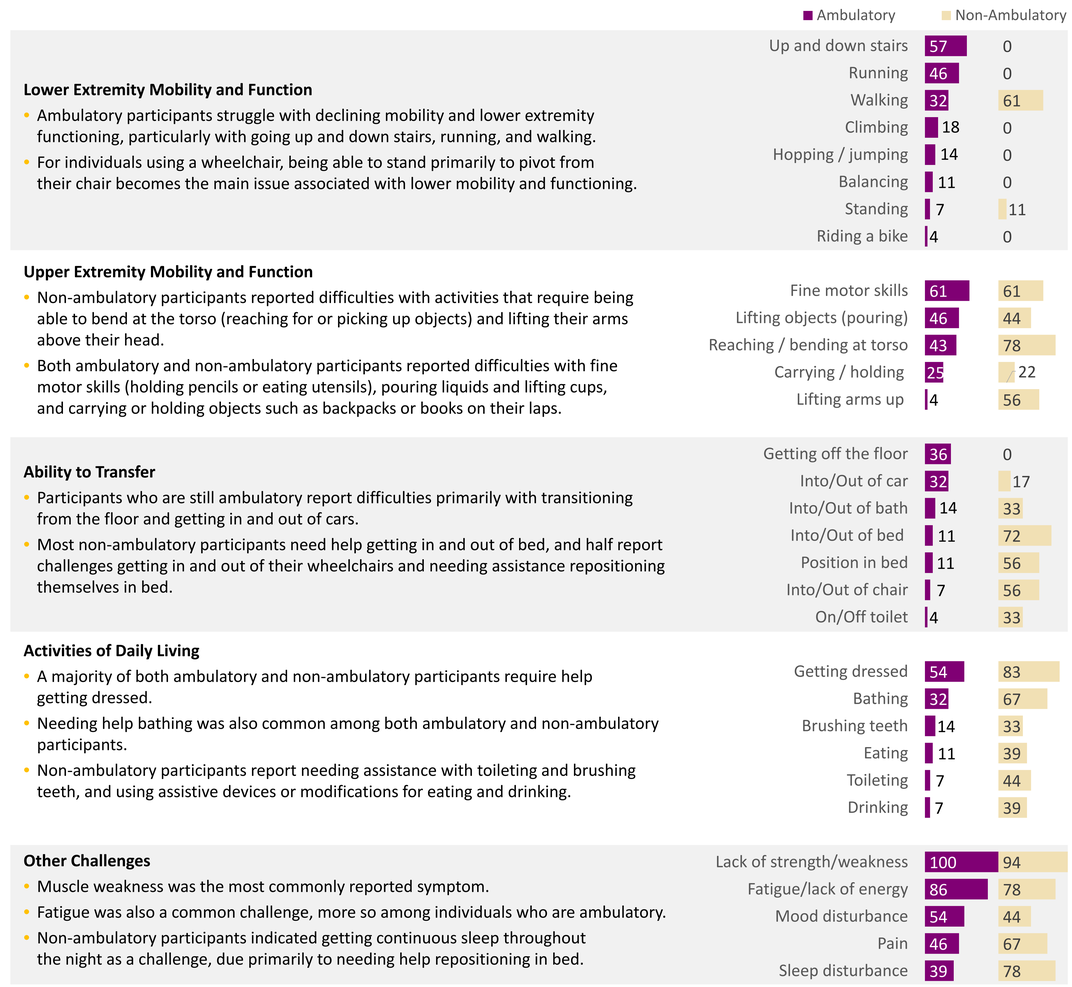
Table 1. Caregiver Characteristics (N=42)

Age of caregiver, y mean (range)	44 (31-78)
Caregiver relationship, n (%)	
Mother	36 (88)
Other	6 (14)
Ethnic origin, n (%)	
White	38 (93)
Other	3 (7)
Education Level, n (%)	
Graduate Degree	13 (32)
College Graduate	8 (20)
Some College	18 (44)
High School/GED	2 (5)
Marital Status, n (%)	
Married	34 (83)
Other	7 (17)
Currently Employed, n (%)	
Caregiver	39 (95)
Spouse	33 (81)
Has Health Insurance, n (%)	26 (63)
Number of children with DMD cared for, mean (range)	1 (1-3)

Key Symptoms and Difficulties of DMD

- Participants were asked to describe impairments they experience in a typical day that are associated within broader domains (e.g. lower mobility, transfers, ADLs). **Figure 1** presents the percent of responses provided by ambulatory and non-ambulatory participants.
- Overall, the most common symptoms and impacts reported across ambulation status related to physical strength (n=45; 98%), transfers (n=34; 74%), activities of daily living (ADLs) (n=40; 87%) and lower mobility and function (n=40; 87%). Fatigue was also commonly reported (n=38; 83%).
- As expected, differences in the specific activities of importance were observed between ambulatory and non-ambulatory participants. Ambulatory dyads identified lower extremity mobility and function issues (e.g. running and climbing up and down stairs). Non-ambulatory dyads identified upper extremity mobility and function issues (e.g., lifting objects and arms up) as well as ability to transfer. Non-ambulatory dyads also reported more issues with ADLs.

Figure 1. Frequency of symptoms and impacts reported by caregivers and patients with DMD by ambulatory status



Lower Extremity Mobility and Function

- Ambulatory participants struggle with declining mobility and lower extremity functioning, particularly with going up and down stairs, running, and walking.
- For individuals using a wheelchair, being able to stand primarily to pivot from their chair becomes the main issue associated with lower mobility and functioning.

Upper Extremity Mobility and Function

- Non-ambulatory participants reported difficulties with activities that require being able to bend at the torso (reaching for or picking up objects) and lifting their arms above their head.
- Both ambulatory and non-ambulatory participants reported difficulties with fine motor skills (holding pencils or eating utensils), pouring liquids and lifting cups, and carrying or holding objects such as backpacks or books on their laps.

Ability to Transfer

- Participants who are still ambulatory report difficulties primarily with transitioning from the floor and getting in and out of cars.
- Most non-ambulatory participants need help getting in and out of bed, and half report challenges getting in and out of their wheelchairs and needing assistance repositioning themselves in bed.

Activities of Daily Living

- A majority of both ambulatory and non-ambulatory participants require help getting dressed.
- Needing help bathing was also common among both ambulatory and non-ambulatory participants.
- Non-ambulatory participants report needing assistance with toileting and brushing teeth, and using assistive devices or modifications for eating and drinking.

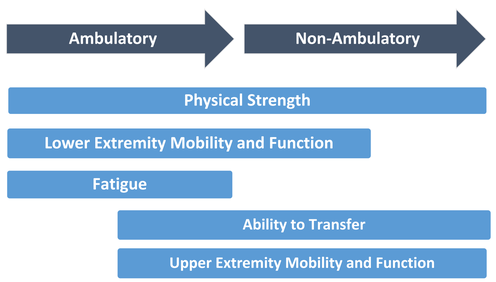
Other Challenges

- Muscle weakness was the most commonly reported symptom.
- Fatigue was also a common challenge, more so among individuals who are ambulatory.
- Non-ambulatory participants indicated getting continuous sleep throughout the night as a challenge, due primarily to needing help repositioning in bed.

Conceptual Model of DMD

- Difficulty with physical strength is experienced throughout all phases of disease progression, whereas difficulty with lower extremity mobility and function is important primarily in early stages of disease progression and until loss of ambulation.
- Fatigue was reported most often in the earlier (ambulatory) phase of disease progression.
- Ability to transfer becomes important as ambulation wanes, and it continues to be important throughout the non-ambulatory phase.
- Upper extremity mobility and function becomes more important in the non-ambulatory phase, although interestingly challenges have also been reported by the ambulatory participants. This may be in part due to some boys being closer to non-ambulatory stage of disease (e.g. transitional) where the use of their hands and arms may be weakened, causing them to have difficulty pouring beverages or carrying books.

Figure 2. Conceptual model of DMD



Biggest Challenges

- **Keeping up with peers** was commonly reported among both ambulatory (n=15; 54%) and non-ambulatory (n=5; 28%) participants.

  I would just think like endurance more like going further and kind of not worrying about his legs wearing out. I think that would be key, yeah. Just kind of keeping up with the kids, that's what I think it would be. [Ambulatory SRPT017, Caregiver]

  So yeah, not being able to walk and play with his friends. That affect his – I mean, that one only – it's affected the most because he cannot keep up with his friends, and sometimes they will be a little mean, so that will have more effect on him. [Non-ambulatory SRPT023, Caregiver]

- **Lower mobility** issues was another common challenge among ambulatory participants (n=14; 50%) and non-ambulatory participants (n=10; 56%).

  It would be like running, that's like the biggest issue. [Ambulatory SRPT104, DMD Individual]

  Able to walk or just a sense of normalcy. I want to be like my brothers, walking, running, playing sports. Yeah, bumping – yeah, all that. [Non-ambulatory SRPT021, DMD Individual]

Treatment Expectations & Benefits

- Ambulatory participants (71%) were more likely to indicate their desire of a new treatment for DMD would be one that would **maintain/stabilize current functioning** than non-ambulatory participants (56%). A third (29%) indicated that that their expectation of a new treatment was one that **slows disease progression**.

  I'd be most looking for stabilization, keep him as close to as he is now. Improvement is fantastic, but in this disease, stabilization is—would be a huge win. [SRPT002, Caregiver]

- Non-ambulatory participants (72%) were more likely to express a desire that a new treatment would lead to **an overall improvement in their symptoms and functioning** than ambulatory participants (61%).

  Like walk—just the strength of muscles, being able to walk, being able to run, just being able to function, you know, prior to the loss. So, that would be the hope. [SRPT013, Caregiver]

- Participants were asked to describe what small changes or improvements from new treatments would make a difference in their lives. For ambulatory participants, **better lower limb mobility** (33%), **improved strength and/or less muscle weakness** (26%), and **improved endurance** (22%) would be the most meaningful benefits.

  Maybe just like giving me more energy. [SRPT016, DMD individual]

- Among non-ambulatory participants, the most frequently reported small but important improvements from a new treatment were **improved strength and/or less muscle weakness** (n=8; 42%) and **greater independence in ADLs** (n=6; 32%).

  More strength for me so I can do more things from my seat. Probably in my legs or in my shoulder area, my arms. [SRPT009, DMD individual]

CONCLUSIONS

- This study contributes to the limited qualitative literature by including both the ambulatory and non-ambulatory patient experience of living with DMD which can inform patient-centered measurement strategies in clinical trials by highlighting the symptoms and impacts that are the most important to DMD individuals across ambulatory spectrum.

REFERENCES

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