# Exploring Quality of Life in Duchenne Muscular Dystrophy: Insights From Qualitative Patient and Caregiver Interviews

Mindy Leffler,[1] Marielle G. Contesse,[1] Eugenio Mercuri,[2] Francina Munell Casadesus,[3] Mercedes Lopez Lobato,[4] Samantha Parker,[5] Sara Cazzaniga,[5] Nicoletta Coceani,[5] Valerie Deroo[5]

[1]Red Nucleus, Yardley, Pennsylvania, USA; [2]Pediatric Neurology Unit, Department of Woman and Child Health and Public Health, Università Cattolica del Sacro Cuore & Centro Clinico Nemo, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, Rome, Italy; [3]Pediatric Neurology, Vall d'Hebron Institut de Recerca, Barcelona, Spain; [4]Pediatric Neurology Unit, Hospital Universitario Virgen del Rocío, Sevilla, Spain; [5]Italfarmaco S.p.A., Milan, Italy

## INTRODUCTION

- Duchenne muscular dystrophy (DMD) is a rare, genetic disease characterized by progressive muscle degeneration and weakness
- As the disease progresses, individuals with DMD become increasingly dependent on caregivers
- While improvements have been made in supportive and rehabilitative care, there is still no cure for DMD
- Because of the progressive nature of DMD, successful treatment may slow functional decline for treated patients
- It is challenging to assess whether the slowing of a patient's functional decline changes the patient's quality of life
- This study explores the impact of both the incremental loss of muscle function and change in ability to perform activities of daily living have on patient and caregiver quality of life

## OBJECTIVES

- To understand the impact of the incremental loss of muscle function on patient and caregiver quality of life
- To understand the impact of changes in the ability to perform activities of daily living on patient and caregiver quality of life

## METHODS

- Qualitative interviews were conducted with US caregivers of individuals with DMD who could no longer raise their hand(s) to their head
- Interviews were conducted remotely using a semistructured interview guide and secure video conferencing platform and lasted approximately 60 minutes
- Interview recordings were transcribed, and deidentified transcripts were single coded for themes using NVivo 15 software
- Results were summarized using representative quotes

## RESULTS

### TABLE 1. Patient age and current level of function

| Caregiver study ID | Patient age, y | Activities the patient can do without any help | Activities the patient requires help with |
|---|---|---|---|
| B01 | 18 | Playing video games and playing on a phone or iPad | Toileting, brushing teeth, scratching anything that is not within limited range of motion |
| B02 | 18 | Playing video games, using phone, and pushing buttons on remote control | Toileting, hygiene, getting dressed, being fed, repositioning at night |
| B03 | 22 | Feeding self, washing parts of themselves in shower, using a computer, playing video games, painting nails | Getting food, taking medicine, getting on toilet, wiping, getting in and out of shower, washing hair, getting things out of backpack, getting desk set up |
| B04 | 23 | Brushing teeth, feeding self | Writing, grooming, dressing, toileting, getting in and out of bed, moving arms in bed, repositioning in bed |
| B05 | 30 | Feeding self, using computer, playing video games, drinking with a straw | Lifting arms to face, repositioning self |
| B06 | 24 | Playing video games, using a mouse, playing board games, drinking bottle of water | Putting blankets on and cutting meat |
| B07 | 22 | Using computer, using phone, pushing up glasses, using wheelchair joystick, writing with pencil or pen | Dressing, toileting, showering, brushing teeth, feeding, lifting a glass |
| B08 | 24 | Scooting, feeding self, using urinal to pee, going to college | Preparing food, dressing, undressing, moving computer around, transferring to shower chair, getting on and off the toilet, showering, washing hair, scratching back, scratching head |

### FIGURE 1. Impact of changes in ability to self-feed on quality of life

**Activity of daily living**

**Feed self**

**Incremental loss**

- **Difficulty using utensils:** cannot use spoons or forks; cannot lift metal utensils; cannot lift runny liquids without spilling
- **Arm and hand limitations:** cannot get hand to mouth; need to support feeding hand; need to prop one hand on the other
- **Positioning needs:** food propped up; lean forward, use table as a lever; precise chair/table setup; cannot eat in car
- **Chewing and fatigue:** gets too tired chewing food; needs food cut up to finger feed

**Equipment required**

- **Adaptive utensils:** lighter utensils, fondue fork
- **Adaptive dishware:** specialized bowls and plates
- **Positioning aids:** book to prop food, higher table, wheelchair table
- **Assistive devices:** robotic arm, manual counterbalanced arm

**Meaningfulness of loss to patient**

- **Social limitations:** meals no longer a social experience; cannot go out with friends; can only eat with supportive friends
- **Public challenges:** avoids eating in public; does not want people staring; stands out among peers
- **Emotional impact:** feelings of depression, shame, and loss of independence

**Impact of changes in ability on caregiver**

- **Caregiving demands:** meals become a caregiving task; cannot be away from child; must feed child ahead of meals
- **Logistical challenges:** more complicated logistics; hard to find equipment; must always plan ahead; must plan who sits next to child
- **Caregiver strain:** increases fatigue

**Caregiver quotes**

- "…he was too embarrassed for my husband to feed him…"
- "We probably would not want to eat in restaurants anymore cause who wants everybody, you know, looking."
- "…at 18 you want your friends to have to feed you…" *(sarcastically)*
- "If he's not able to eat, he does not want to live."
- "It changes the way you sit at a table, like you have to be in a certain position so you can feed him…"

### FIGURE 2. Abilities that depend on arm function, as reported by caregivers

| Muscle function | Arm function | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| Muscle function sub-category | Raise hand to face | | | | | | Raise hand to mouth | Engage arm muscles | Lift forearms off lap | Lock out elbows to push off | | Lateral arm movement | Reach | Lift weight |
| Abilities that depend on muscle function | Scratch itch | Protect face | Get hair out of mouth | Push glasses up | Brush teeth | Have a phone conversation not on speaker | Self-feed | Get dressed | Get dressed | Get dressed | Transfer | Drive in adaptive car | Use full keyboard | Use phone |
| Impact on patient and caregiver (representative quotes) | "He'll get mosquito bites and…he can't scratch it." | "…he wouldn't be able to put his hands up and protect his face or turn his head in time." | "Imagine having a hair in your mouth you can't get it out, you have to rely on somebody else to find it, get it for you. I was just like; this disease is relentless." | "I just worry, is somebody gonna be able to push his glasses up, you know, that little thing, if he needs it." | "He cannot brush his teeth himself." | "You have no privacy whatsoever anyway, and then you can't even talk to your friends without being on a speakerphone." | "Being fed a meal as a…17/18-year-old kid is kind of humiliating." | "It's like putting a pillowcase on the flattest, limpest pillow you could ever have." | "So that small bit of help that he could do with helping to get a shirt on…to be able to lift his, his hands, his forearms up a bit. He can't do that anymore." | "He would scoot…his bum up a little bit so that I can pull on underwear or shorts or pants. and that was his arm muscles." | "He used to be able to scooch forward on his bed, he can't do that." | "He started cornering much more slowly out of fear that if…his elbow fell off his armrest, he wouldn't be able to get it back up and be able to drive." | "…he can reach the lower couple of rows of keys, but he can't reach the upper row with the numbers." | "He cannot lift that anymore… we had to work around it…my arms are now his arms." |

## CONCLUSIONS

- Independent activities of daily living for patients with DMD undergo a qualitative degeneration comprised of incremental and meaningful losses in function
- There are a number of impactful and meaningful functional losses in patients with DMD that affect quality of life and are not captured in clinical trial outcomes
- Capturing and quantifying the nuances of loss in patients with DMD will provide a more complete picture of the function of people with DMD for regulators and payers
- As part of this study, additional qualitative interviews are currently being conducted with patients treated with givinostat and their caregivers. Future analyses will explore the impact of the maintenance of independence on quality of life once all interviews have been conducted

**ACKNOWLEDGMENT AND FUNDING**

This study is sponsored by Italfarmaco S.p.A.
Editorial support was provided by Jennifer Ayala, PhD, CMPP, from Citrus Health Group, Inc. (Chicago, Illinois), and was funded by ITF Therapeutics, LLC.

**AUTHOR DISCLOSURES**

**ML and MGC:** Employees of Red Nucleus.
**EM:** Received grants or research support from Astellas Pharma, BioMarin Pharmaceutical, Capricor Therapeutics, Catabasis Pharmaceuticals, Edgewise Therapeutics, Italfarmaco S.p.A., Pfizer, PTC Therapeutics, and Santhera Pharmaceuticals and consulting fees from Sarepta Therapeutics, Astellas Pharma, Avidity Biosciences, BioMarin Pharmaceutical, Bristol Myers Squibb, Capricor Therapeutics, Catabasis Pharmaceuticals, Edgewise Therapeutics, Eli Lilly, Epirium Bio, Entrada Therapeutics, Gilead Sciences, Halo Therapeutics, Italfarmaco S.p.A., Novartis, PepGen, Pfizer, PTC Therapeutics, Prosensa, and Santhera Pharmaceuticals.
**FMC:** Principal investigator of the Vall d'Hebron University Hospital, Barcelona, site of the current study. **MLL:** Received research support from Italfarmaco S.p.A. **SP, SC, NC, and VD:** Employees of Italfarmaco S.p.A.

ITALFARMACO

Presented at ISPOR Europe 2025 | 9–12 November 2025 | Glasgow, Scotland, UK