

# Qualitative interviews with caregivers of children aged 5–7 years with Duchenne muscular dystrophy to assess content validity of the proxy-completed DMD-QOL

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## Introduction

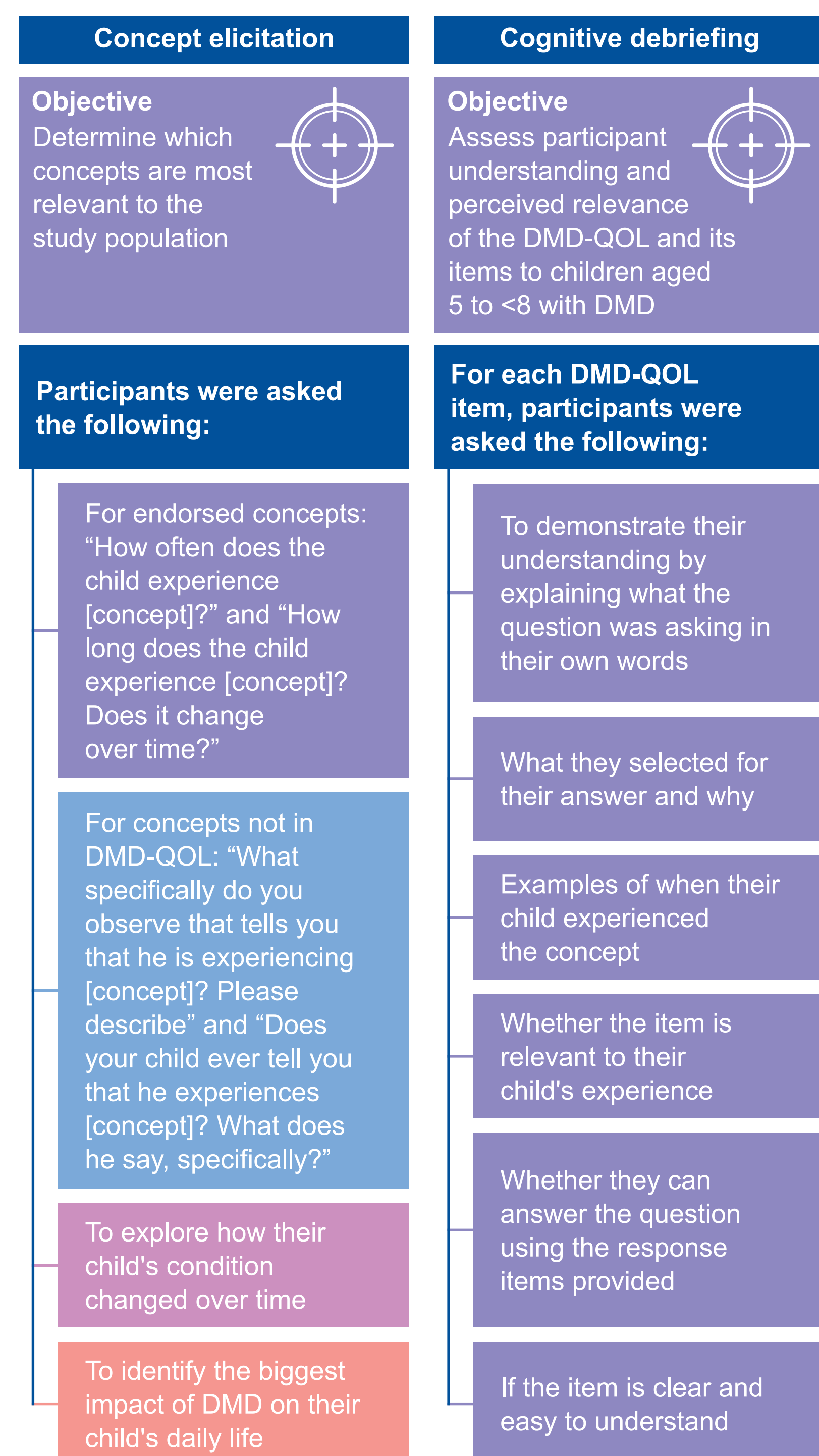
- Duchenne muscular dystrophy (DMD) is a severe X-linked neuromuscular disorder that causes progressive muscle weakness, cardiopulmonary impairments, and behavioral symptoms that significantly impact quality of life (QOL)<sup>1–3</sup>
- Clinical symptoms of DMD emerge at approximately 3 years of age, whereas definitive diagnosis most often occurs between 3 and 5 years of age<sup>4</sup>
- Clinical trials of DMD commonly enroll participants as young as 4 years of age
- The DMD-QOL questionnaire is a 14-item questionnaire designed to assess physical functioning, psychological impact, and social participation in males with DMD
- However, DMD-QOL was validated for proxy reporting in boys starting at 7 years of age and by proxy or self-report starting at 10 years of age, leaving an evidence gap for younger children

## Objectives

- To assess the content validity of proxy-completed DMD-QOL for children 5 to <8 years of age by
  - Determining if the DMD-QOL aligns with caregiver perceptions, child physical functioning, psychological impact, and social participation associated with DMD
  - Assessing the relevance, comprehensiveness, ease of use, and understandability of the proxy-completed DMD-QOL

## Methods

- This was a cross-sectional, non-interventional qualitative study that conducted 1-on-1 hybrid concept elicitation and cognitive debriefing interviews via telephone or teleconference
- Eligible participants were adult caregivers of children with DMD aged 5 to <8 years



### Interim results from the first 5 interviews were assessed and discussed with the study sponsor and advisors before conducting the remaining 4 interviews

- Based on the first 5 interviews, it was not clear to participants whether to respond to items based on their child's experience in general or experiences related to DMD
- For the final 4 interviews, participants were shown the instruction, "For the following questionnaire, please respond about impacts your child, or the person you are responding on behalf of, may experience related to their Duchenne Muscular Dystrophy (DMD)," to see if it would impact their interpretation or responses
- This additional instruction was only shown to participants at the end of their interview to ensure it did not influence their initial feedback about the DMD-QOL

## Results

### Demographics

- All participants were parents of children with DMD; most were female and had a post-secondary degree (Table 1)
- Children with DMD were male and 6.5 years of age on average at the time of interview; 6 were from the US, and 3 were from the UK (Table 2)

**Table 1. Caregiver characteristics and demographics**

| Characteristic, n (%)               | Caregivers (N = 9) |
|-------------------------------------|--------------------|
| <b>Number of children with DMD</b>  |                    |
| 1                                   | 8 (89%)            |
| 2                                   | 1 (11%)            |
| <b>Female</b>                       | 7 (78%)            |
| <b>Post-secondary degree</b>        | 6 (67%)            |
| <b>Employment</b>                   |                    |
| Employed full-time                  | 4 (44%)            |
| Employed part-time                  | 2 (22%)            |
| Not employed outside the home       | 3 (33%)            |
| <b>Annual household income (US)</b> |                    |
| \$100,000–\$149,999                 | 2 (22%)            |
| \$50,000–\$74,999                   | 2 (22%)            |
| Decline to answer                   | 2 (22%)            |
| <b>Annual household income (UK)</b> |                    |
| ≥£150,000                           | 1 (11%)            |
| £100,000–£149,999                   | 1 (11%)            |
| £35,000–£49,999                     | 1 (11%)            |

DMD, Duchenne muscular dystrophy.

**Table 2. Characteristics and demographics of children with DMD**

| Characteristic                         |                           |
|--|---------------------------|
| <b>Age at time of interview, years</b> |                           |
| Mean (SD); median (min, max) in US     | 6.9 (1.0); 7.2 (5.7, 7.9) |
| Mean (SD); median (min, max) in UK     | 5.7 (0.7); 5.7 (5.1, 6.4) |
| <b>Age at diagnosis, years</b>         |                           |
| Mean in US                             | 3.3                       |
| Mean in UK                             | 4.6                       |
| <b>Ethnicity (US)</b>                  | N = 6                     |
| White and not Hispanic, n (%)          | 6 (100%)                  |
| <b>Ethnicity (UK)</b>                  | N = 3                     |
| Multiple ethnic groups, n (%)          | 1 (33%)                   |
| Black (Caribbean) British, n (%)       | 1 (33%)                   |
| White, n (%)                           | 1 (33%)                   |

DMD, Duchenne muscular dystrophy; max, maximum; min, minimum; SD, standard deviation.

### Concept elicitation

- The most frequently endorsed concept domains were related to fatigue, mobility, and psychosocial impacts

- All participants (9/9; 100%) said their child felt tired
- Most participants (8/9; 89%) said their child had difficulty getting around
- Most participants (7/9; 78%) said their child felt angry
- Most participants (7/9; 78%) said their child couldn't participate in all activities with friends
- Most participants (6/9; 67%) said their child couldn't do all the things he wanted to

### Cognitive debriefing

- Overall, participants found the questionnaire comprehensive and clear

Six participants (66.7%) did not suggest changes to the instructions; of the remaining 3 (33.3%; responses not mutually exclusive), 2 suggested clarification that questions are about DMD rather than life in general, 1 suggested extending the recall period, and 1 suggested asking respondents to specify if the experienced impacts are due to difficulty rather than avoidance

Some participants found it difficult to choose an answer because their child experienced the impact unrelated to DMD

**"The only one where I [had difficulty was], oh, number four, he was in pain. He was in pain, but it wasn't because of DMD. Like, he broke his wrist, which was unrelated"**

- Participants said that some items were not relevant to their child now but may be in the future and that some were not relevant to their child but may be for other children (Table 3)

**Table 3. Relevance of concepts**

| Concept domains                               | Relevant n (%) |
|---|----------------|
| He found it hard to get around                | 9 (100%)       |
| He felt tired                                 | 8 (89%)        |
| He found it hard to use his hands             | 7 (78%)        |
| He was in pain                                | 7 (78%)        |
| He could take part in things with his friends | 7 (78%)        |
| He felt angry                                 | 7 (78%)        |
| He felt unhappy                               | 6 (67%)        |
| He could take part in the things he wanted to | 6 (67%)        |
| He felt worried                               | 5 (56%)        |
| He felt good about himself                    | 5 (56%)        |
| He found it hard to talk to people            | 4 (44%)        |
| He felt embarrassed                           | 4 (44%)        |
| He found it hard to breathe                   | 3 (33%)        |
| He found it hard to eat                       | 2 (22%)        |

DMD, Duchenne muscular dystrophy.

- Ten of the 14 DMD-QOL items were endorsed as relevant by a majority of respondents. All items were endorsed as relevant by at least 1 participant
- For 8 of the 14 DMD-QOL items, all caregivers who endorsed the concept item as relevant were able to directly observe their child experiencing the impact (Table 4)

**Table 4. Source of information for DMD-QOL items**

| DMD-QOL item   | Participants endorsing (n) | Direct observation (n) | Child report (n) | Third-party/other report (n) |
|--|----------------------------|------------------------|------------------|------------------------------|
| He found it hard to use his hands                          | 7                          | 7                      | 0                | 0                            |
| He found it hard to eat                                    | 2                          | 2                      | 0                | 0                            |
| He found it hard to breathe                                | 2                          | 2                      | 0                | 0                            |
| He was in pain <sup>a</sup>                                | 7                          | 6                      | 1                | 0                            |
| He felt tired  | 9                          | 9                      | 0                | 0                            |
| He found it hard to talk to people                         | 5                          | 5                      | 0                | 0                            |
| He felt good about himself <sup>a</sup>                    | 9                          | 8                      | 1                | 0                            |
| He felt unhappy  | 8                          | 8                      | 0                | 0                            |
| He felt embarrassed <sup>a</sup>                           | 7                          | 5                      | 2                | 0                            |
| He felt worried <sup>a</sup>                               | 8                          | 5                      | 3                | 0                            |
| He felt angry  | 8                          | 8                      | 0                | 0                            |
| He found it hard to get around                             | 8                          | 7                      | 1                | 0                            |
| He could take part in the things he wanted to <sup>a</sup> | 5                          | 3                      | 1                | 1                            |
| He could take part in things with his friends              | 6                          | 5                      | 1                | 0                            |

<sup>a</sup>Item for which at least 1 caregiver relied on child report or third-party report to answer. DMD-QOL, Duchenne muscular dystrophy quality of life questionnaire.

### Perceptions of meaningful treatment benefit

- All participants (9/9; 100%) said it would be meaningful if the impacts of their child's DMD remain the same over time
- All participants (9/9; 100%) said slowing of disease worsening would be a meaningful treatment benefit

"Just knowing what is to come, like if everything just stayed the same, as it is, like that's easy. We can handle that. Later things are just unknown and scary"

"If the deterioration can be slowed, stabilized for as long as possible to avoid, or not avoid, but delay needing to go into a wheelchair, that would be absolutely amazing"

"So like right now, I feel like he is the healthiest he'll ever be with this disease...knowing the way that this can progress, right now he really is in a good place where he's not — like I know there are kids his age who already need the wheelchair for way more"

"...the nature of this condition actually get worse...if I got that kind of treatment that is going to maybe stable him...not going to make him get worse than how it is...I would appreciate it"

"So if I had to rank them, like obviously we would want progress, that would be preferred. Stopping it so that it didn't get worse would be the second choice, and then the third choice would be just at a slower rate. So I would accept that as well"

"...that would be the aim, as slow as possible...the main things we are looking for, quality of life and the extension of life than if he is ambulatory, non-ambulatory, it's something that we can deal with"

"...if you can delay the deterioration to get to the next helpful medication, that's a good thing generally, because you cannot usually get back what you have lost. But if something similar comes along in, say, two years that he can't participate in [because the first treatment limited his ability to participate in other future treatments]...it's hard"

"I think I'd have to know some of the parameters around that and what some of the side effects are. [There are]...treatments that...we have chosen not to use because of side effects...if those side effects would outweigh the decrease of progression, then I would have to consider it in light of that"

## Conclusions

- Overall caregiver feedback indicated that the proxy-completed DMD-QOL is easy to use, understandable, and face valid for use by caregivers of children aged 5–7 years with DMD
- All participants were able to complete the questionnaire on their child's behalf, and 10 of the 14 DMD-QOL items were endorsed by a majority of respondents as relevant to their child
- Certain DMD-QOL concepts, such as feeling worried, embarrassed, or angry, may be of variable relevance and observability to participants, potentially due to their child's stage of psychosocial development or disease progression
- Additional concepts, such as finding it hard to talk to people, feeling embarrassed, and finding it hard to eat or breathe, were less frequently reported as relevant. However, all DMD-QOL concepts were considered relevant by at least 1 interview participant
- Additional research may be warranted to confirm generalizability of study findings and to further evaluate the utility of DMD-QOL with younger populations, including among children aged 4 years, in clinical studies

### References

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### Disclosures

AP, EC, MC, and ED-A are employees and shareholders of BioMarin Pharmaceutical Inc. PP and JC are developers of the DMD-QOL and have received research funding from Duchenne UK. KG, EB, and HG are employees and shareholders of Evidera | PPT, which was contracted by BioMarin Pharmaceutical Inc. to conduct the described study. This research was previously presented at the 30th annual International Congress of the World Muscle Society (WMS) on October 7–11, 2025, in Vienna, Austria.

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