

Development of a model of the patient experience of Becker Muscular Dystrophy (BMD)

A literature review and qualitative interview study

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Background

- Becker muscular dystrophy (BMD) is a rare progressive neuromuscular disorder, resulting from mutations in the DMD gene on the X chromosome, which codes for the protein dystrophin (1).
- BMD has a substantial impact on health-related quality of life for patients and associated disease burden, however little research has been conducted to fully understand disease burden and unmet need (2, 3).
- Understanding the patient experience of BMD is important for selecting or developing patient reported outcome measures (PROs) – questionnaires or surveys that are completed by patients, for example in clinical trials, to report how their condition is affecting their health-related quality of life.

Objectives

- This study aimed to develop a model of the adult and pediatric experience of BMD, including disease symptoms and impact on functioning and wellbeing, via completion of a literature review and qualitative interview study.

Methods

- A literature review was conducted to identify relevant qualitative studies or patient testimonials from journal articles, conference proceedings and patient advocacy group websites.
- Interviews lasting 45 minutes were conducted with patients with BMD aged 12–55 years (and/or their caregivers). Participants were from the US.
- Inclusion criteria required patients to be ambulatory but could have a range of disease severity.
- Interviews used open-ended questions to explore patient experiences of life with BMD, including the symptoms, and impacts that they experience.
- Participants rated each symptom they experienced on a scale of 0–10 based on “bothersomeness”, i.e. how much does the symptom bother them when it is at its worst.
- Patients were also asked to rank their top 5 most bothersome impacts of living with BMD.
- Data were analyzed using thematic analysis techniques, and a model was developed to depict the experience of BMD.

Results

Literature review

One journal article (4) and one patient testimonial (5) were identified in the literature review. Pain, fatigue, leg cramping and impacts on activities of daily living (ADLs) and physical functioning were identified as important concepts.

Qualitative study

Demographics

15 interviews were conducted:

- n=8 interviews with patients aged 18–55;
- n=5 dyad interviews with patients aged 12–17 and their caregivers;
- n=2 interviews with caregivers of patients aged 12–17.

Patients with varying disease severity were interviewed; this was defined according to ambulatory status:

- AM1: Fully ambulatory, no walking aid or help required, n=7;
- AM2: Ambulatory, but walking aid or help required sometimes/most of the time, n=6;
- AM3: Ambulatory, but always need to use a walking aid or have help to walk, n=2.

Most patients were receiving, or had previously received, treatment for their BMD, including:

- Physical therapy, n=10;
- Heart/blood pressure/diuretic medication, n=5;
- Muscle relaxers, n=1;
- Steroids, n=1;
- Muscle stretching and lengthening, n=1;
- Two patients had never received any treatment for their BMD.

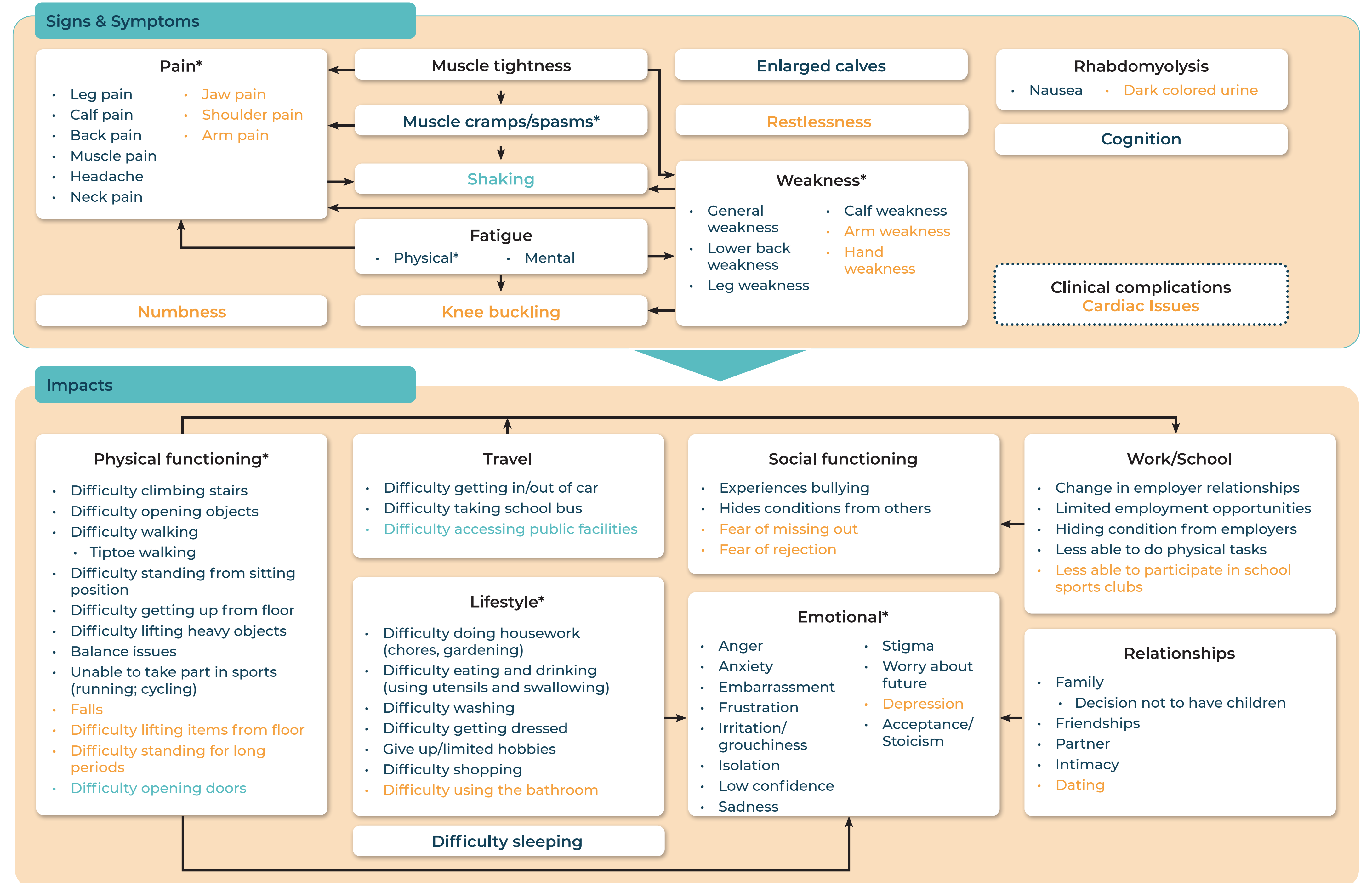
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Model

A model of the patient experience of BMD is presented in Figure 1.

Figure 1. Model describing the patient experience of BMD - A conceptual model of the patient experience of Becker Muscular Dystrophy (delineated by severity)

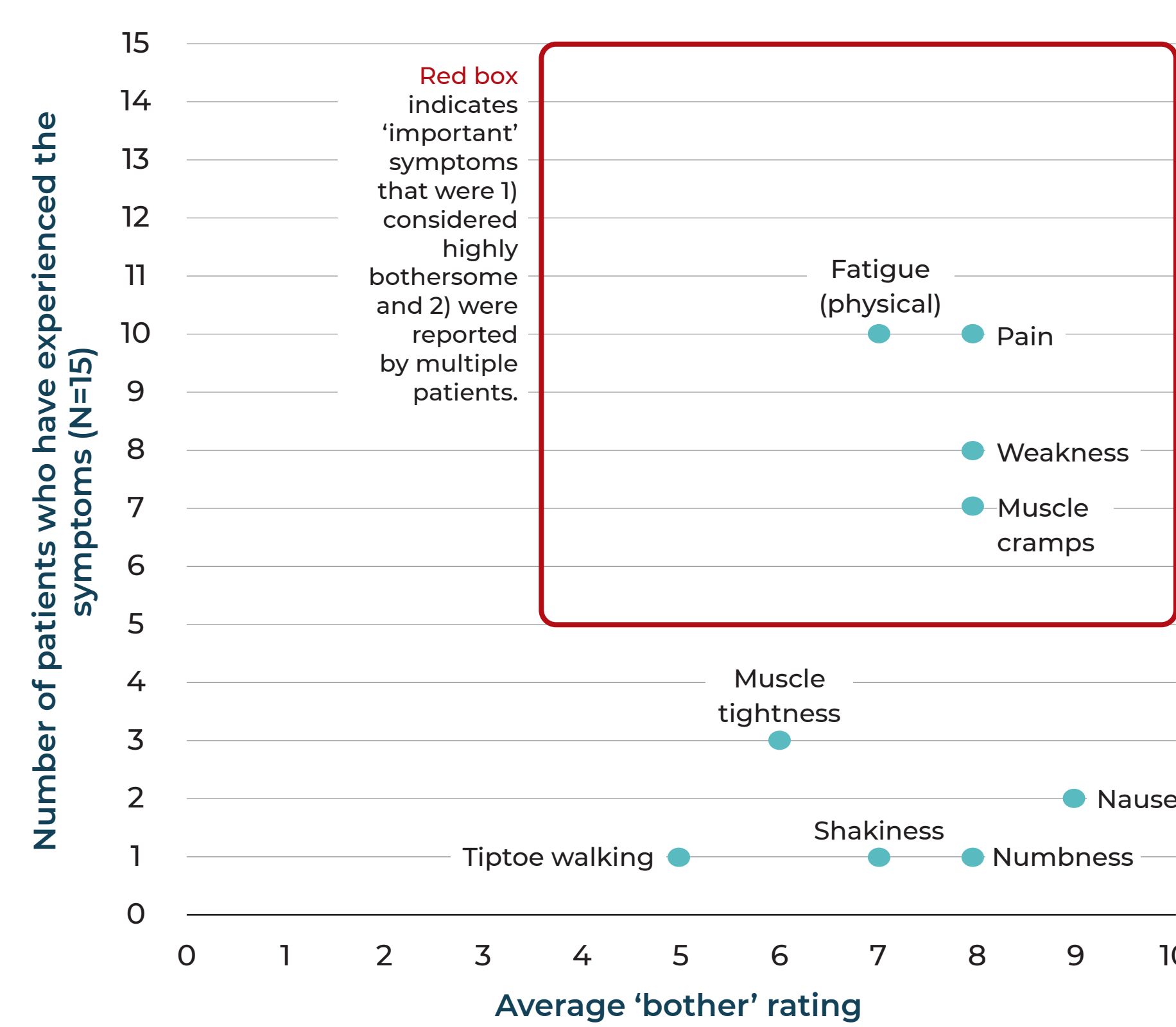


Key
→ Arrows indicate posited relationships between domains;
Domains/concepts in navy font start at AM1 severity (fully ambulatory, no walking aid/assistance required)
Domains/concepts in orange font start at AM2 severity (ambulatory, with walking aid/assistance used sometimes)
Domains/concepts in blue font start at AM3 severity (ambulatory, with walking aid/assistance used most of the time or always)
* Asterisk indicate salient domains/concepts reported by patients and/or caregivers
..... Concepts in a dashed box represent parts of the patient experience that are unlikely to be relevant for patient-reported measurement in a clinical trial

Important symptoms

- Figure 2 displays participants' average 'bothersomeness' scores for each symptom and the number of people who experienced each symptom. Symptoms that were rated as highly bothersome and that were experienced by multiple patients were considered to be 'important'.
- Physical fatigue, pain, weakness, and muscle cramps were considered to be important to the patient experience.
- Other symptoms (e.g., numbness and nausea) ranked high in disturbance rating but were only reported by one or two participants.

Figure 2. Important BMD symptoms



“If you're in a lot of pain and, you know, just don't want to think about anything, but just, like, being in pain...like, you can't even figure how to help yourself because you're just hurting so much. And you're just, like, wanting to sit there, trying to get a sleep, so you don't have to feel it that way.” (Adult, AM1)

“When it's at its worst, [weakness] severely limits me doing anything. Err, either I have to rest or I'm just not going to do anything else and it's very limiting.” (Adult, AM3)

“At its worst it was cramping, um, that wouldn't go away, whether it was, kind of, massaging it or provide heat to it. Um, it would, it would last forever, and it was, um an, an excruciating pain that just, you know, you – one of those situations where it made – makes your eyes water and, you know, you just, you don't want to move.” (Adult, AM2)

“I mean, [fatigue] just feels like you don't want to get up. Like, even if you need to make food or something, you're just, like, no, like, I'm just going to stay in bed all day. Like, you don't want to have to put weight on anything. You just want to sit there and, like, watch TV and do nothing. I guess that's, like, the best way to explain it.” (Adult, AM1)

Most bothersome impacts

| Domain (n*) | Quote |
|----------------------------------|---|
| Physical mobility and ADLs (n=9) | “Um, probably the most bothersome is just not being able to be as active as he would like to be. Err, you know, ‘cause like I said, he – err, as a child, he was very into sports, err, you know, bike riding, skateboarding, err, soccer, all those types of things.” (Caregiver, AM1) |
| Emotional (n=7) | “I don't know what to classify this as, but maybe, like, my own, like, mental health impacts of, like, dealing with all of the constant questions and unknowns.” (Adult, AM1) |
| Social (n=3) | “The societal impact of, like, when I do tell people, it's like “What is that?” and “How – what are you going to do?” Err, you know, just the questions from the other end.” (Adult, AM1) |
| Work (n=2) | “I would say I feel like eventually it will be my employment, occupational impact, in that, you know, 20/30 years from now, am I going to be, you know, as physical in my work, or not? (Adult, AM1) |
| Relationships (n=2) | “Um, most bothersome I would say is the, the mental relationships. Err, well, you know, the, the, the, the mental effects, err, um, that, you know, the disease has on me and how it affects relationships.” (Adult, AM2) |

*Total number of participants who mentioned at least one impact on this aspect of their life as one of the top 5 most bothersome impacts of living with BMD.

Conclusions

- The newly developed model describes the adult and pediatric experience of BMD.
- Pain, fatigue, weakness, and muscle cramps/spasms are important symptoms, and impacts on physical functioning and emotional wellbeing are most bothersome/important to patients.
- Many symptoms and impacts were identified, highlighting the diverse experience of BMD patients and the unmet need of the condition.
- The model will inform the identification of PRO measures to assess the signs, symptoms, and impacts of BMD, ensuring they are relevant and important to the patient experience.
- Future interviews will also help to refine the model:
 - While pain and fatigue were highlighted as important symptoms, little about their impacts was learned;
 - Some of the symptoms identified were described by only a small number of patients and as yet their relevance/importance are not fully understood (e.g., shaking, knee buckling, restlessness, and others);
 - The model differentiates between concepts experienced by patients of different ambulatory statuses but is based on small numbers of patients. This differentiation needs to be confirmed.
- Future interviews are planned with N=15 patients/caregivers to debrief PRO measures and confirm they are appropriate for use with people with BMD. Collectively, these activities will inform the development of a patient-focused outcome measurement strategy for use in future BMD clinical trials.