Development of a conceptual model of the patient experience of Becker Muscular Dystrophy (BMD) 
A literature review and qualitative interview study

1. Background

- Becker muscular dystrophy (BMD) is a rare progressive neuromuscular disorder resulting from mutations in the X chromosome, which codes for the protein dystrophin.
- 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 the adult and pediatric experience of BMD.
- Understanding the patient experience of BMD is important for selecting and 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.

2. Objectives

This study aimed to develop a conceptual 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.

3. Methods

- A literature review was conducted to identify relevant qualitative studies or 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 18–50,
    - 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=4.

4. Results

Literature Review

One journal article (6) 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

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4. Results (Continued)

<table>
<thead>
<tr>
<th>Signs &amp; Symptoms</th>
<th>Muscle tightness</th>
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<tbody>
<tr>
<td>Pain*</td>
<td>Muscle cramps/spasms*</td>
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<tr>
<td>Muscle loss</td>
<td>Numbness</td>
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<tr>
<td>Fatigue</td>
<td>Knee Buckling</td>
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<td>Difficulty standing</td>
<td>Headache</td>
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Figure 1. Conceptual model describing the patient experience of BMD - A conceptual model of the patient experience of BMD (delineated by severity)

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<thead>
<tr>
<th>Impacts</th>
<th>Travel</th>
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<td>Difficulty getting in/out of car</td>
<td>Difficulty accessing public facilities</td>
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<th>Social functioning</th>
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<td>Experience building</td>
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<th>Emotional*</th>
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<th>Relationships</th>
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<td>Partner</td>
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5. Conclusions

- The newly developed conceptual model describes the adult and pediatric experience of BMD.
- Pain, fatigue, weakness, and muscle cramps are important symptoms, and impacts on physical functioning and emotional wellbeing are most botherome/important to patients.
- Many symptoms and impacts were identified, highlighting the diverse experience of BMD patients and the extent of the need for condition.
- The conceptual model will inform the identification of PRO measures to assess the symptoms, impacts, and quality of life measures, ensuring they are relevant and important to the patient experience.
- Future interviews will also help to refine the conceptual 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 conceptual model differentiates between concepts experienced by patients of different ambulatory statuses but is based on small numbers of patients. This differentiation needs to be improved.

Future interviews are planned with N=15 patients/caregivers to define 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.

References
