

1. Objective

The objective of this study was to develop a strategy to mobilize the Becker muscular dystrophy (Becker) community by capturing and communicating the practical and psycho-social experiences, needs, and challenges of individuals living with Becker and their caregivers across their lifespan.

2. Background

Becker is a serious, debilitating, and potentially life-threatening neuromuscular disorder with a high unmet medical need. The community is underserved as there are:

- No approved therapies for Becker
- Only two interventional trials are actively recruiting, demonstrating the limited research exclusively dedicated to this indication
- No Becker-specific advocacy organizations to promote research and provide patient -focused resources.

Increasingly, the FDA encourages the patient voice to be included in drug development, to help establish the therapeutic context for risk/benefit decision making, improve study design, and enhance trial enrollment and communications. (1,2) Understanding the patient voice is crucial to effective communications and community mobilization, particularly in a community that is underserved and disenfranchised.

3. Design

The design of the patient experience mapping (PEM) study consisted of five stages, with the final community building stage still ongoing (Figure 1). Demographics show that the 14 individuals who participated represented a wide range of age and ambulatory ability. (Table 1) Interviews focused on the journey from pre-diagnosis to diagnosis to treatment and management of Becker.

Table 1: Demographics of interview participants.

	Age range (yrs.)	Range of age at dx (yrs.)	Fully ambulatory	Requires mobility aid	FT Wheelchair
Pediatric n = 7	8-17	2-9	4	3	0
Adult n = 7	30-62	5-30	1	2	4

3. Design (Continued)

Figure 1: Methodology.

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graph LR; A[Patient Advocacy Group (PAG) Interviews] --> B[PEM Recruitment]; B --> C[Qualitative Interviews]; C --> D[Validate & Report]; D --> E[Community Building]
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Patient Advocacy Group (PAG) Interviews	PEM Recruitment	Qualitative Interviews	Validate & Report	Community Building
<ul style="list-style-type: none">Identified 10 patient and multi-stakeholder organizations in Canada, Italy, UK, USFacilitated interviews with five organizations based on group availability, capacity, and interest in BMD space	<ul style="list-style-type: none">Screening survey distributed via muscular dystrophy and Becker-specific social media groupsScreening benchmarks:<ul style="list-style-type: none">US geographyConfirmed BMD diagnosisAmbulation and assistive devices*Years with diseaseAgeRace & ethnicity	<p>One-on-one virtual qualitative interviews were conducted with 14 patients and caregivers.</p> <p>Key topics covered across the journey included pre-diagnosis, diagnosis, treatment, and disease management.</p>	<p>Cross-functional read-out & discussion</p> <p>Advisory Board of select patients/caregivers to validate maps and expand PEM insights/findings</p> <p>Report Back to Key PAG: CureDuchenne, MDA, and PPMD</p>	<p>"Did You Know: Social Media Campaign</p> <p>"It's Time to Get Real About Becker" Educational Campaign</p> <p>Becker Awareness Days at Select Sites</p> <p>Collaborations with Research Organizations and Advocacy Organizations</p>

**participants were selected with the intent to represent a wide range of experiences and stages of disease*

4. Results

Results of the PEM study yielded distinct themes further characterizing the misunderstood and underserved nature of Becker. Diagnostic delays were noted (Figure 2) along with the repercussion of receiving a Becker versus Duchenne diagnosis (Figure 3). Along with mobility limitations,, all participants reported comorbidities with cardiac complications being most concerning. All noted long-term mental health impacts, partially driven by financial worries, that come with living with a disease that has an unknown progression. (Figure 4). While these challenges and experiences demonstrated the significant impact of living with Becker, resources and information about Becker are lacking in the community often leaving these individuals feeling unheard, unseen and under resourced.

Highlights of PEM Results

Figure 2: Diagnosis Insights.

The infographic is titled "DIAGNOSIS" in large, bold, black letters. Below the title, there are two main sections: "KEY FINDINGS" and "COMMUNITY PAIN POINTS".

KEY FINDINGS is divided into three columns, each with an icon and a text box:

- Left:** An icon of a staircase with an arrow pointing up. Text: "Difficulty navigating stairs, running, and falling were common drivers for seeking medical care".
- Middle:** An icon of two feet. Text: "Common physical symptoms noticed by parents included fatigue, enlarged or tight calves, and 'toe walking'".
- Right:** An icon of a head with a brain and a magnifying glass. Text: "All reported a delayed diagnosis with access to a neurologist, geneticist, or other specialist being key to getting a proper diagnosis".

A quote from a caregiver is placed below the key findings, connected by lines to the "KEY FINDINGS" and "COMMUNITY PAIN POINTS" sections:

"We were not being listened to for **nearly 7 years**, always people thought he was doing it for attention... on top of it all, in a clinical setting they weren't seeing the key symptoms within their exams. It made me feel like it was all in my head. Finally, his teacher said something was not right, and her additional advocacy helped us get through to our pediatrician." – caregiver to individual living with BMD

COMMUNITY PAIN POINTS is a section with a list of three bullet points:

- Lack of information/education for primary care providers (i.e., pediatrics, family medicine, and adult internal medicine) on the signs and symptoms of muscular dystrophies means accessing a specialist is key
- Once diagnosed the absence of Becker-specific community for support leaves patients with little resources and information to navigate the complex care required to manage the disease

4. Results (Continued)

Figure 3: A Becker versus Duchenne diagnosis impacts provider communication and access.

A BECKER VERSUS DUCHENNE DIAGNOSIS IMPACTS PROVIDER COMMUNICATION AND ACCESS

BMD is commonly described as the “lesser” or “better” form of Duchenne

- The heterogeneity of BMD leaves patients and caregivers with uncertainty regarding progression and the future, yet the diagnosis is often delivered with more hope and positive outlook than DMD, which can discount the experience of patients and caregivers faced with a life-changing diagnosis

Lack of general public awareness about types of muscular dystrophies

- Patients feel misunderstood and are often labeled as having DMD (in childhood) or multiple sclerosis (in adulthood)
- Lay-public and non-specialist providers are often unaware that there are different types of muscular dystrophy, and are less aware of BMD comparatively to DMD

Resources are often dependent on diagnosis, not symptom severity

- Access to Medicaid coverage can be denied to BMD patients because their diagnosis is “not severe enough” compared to Duchenne
- Patients and caregivers find that patient advocacy group resources are tailored to the Duchenne community

Figure 4: Overall impact of Becker

OVERALL IMPACT OF BMD

KEY FINDINGS

- All participants expressed that living with BMD has had an effect on their mental health and those around them
- Financial impacts were reported across interviews including navigating insurance, unemployment, and home accessibility
- Careful consideration and planning becomes part of daily life and impacts QoL activities like travel and family activities

“I was told you’re lucky you don’t have DMD, and yes, I feel bad for them, but also frustrating that you live longer and live to be 60 or 70 years old, you are constantly going downhill. You have to learn to live with this long-term and the financial aspects that go a long with that. I have to be constantly adjusting my life and you just don’t know what the future holds.” – individual living with BMD

COMMUNITY PAIN POINTS

- Financial burden has a huge negative impact on patient and family mental health and quality of life
- Unknown rate of progression makes long-term planning difficult for housing, etc.
- Mental health struggles stem from various causes but affect multiple areas of patient and family life

Awareness & Education Campaign

From these results, a multi-pronged, multi-stakeholder awareness and education campaign was developed with three goals:

- Assure the Becker community that their unique needs are being heard
- Provide much needed information on Becker
- Encourage the community to come together to create change.

The campaign consisted of articles/flyers disseminated through various channels, (Figure 5) and a series of vignettes on social media focusing on impacts identified in the qualitative research (Figure 6).

The intent of the campaign is to assure the Becker community that their unique needs are being heard, provide much needed information on Becker and encourage the community to come together to create change.

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5. Discussion

These patient experience results show that qualitative data can be powerful and should not be considered anecdotal. When the patient voice and disease misperceptions are visually and emotively communicated, all stakeholders in an underserved community will engage in efforts to serve the community. The lived experiences of the Becker community must be communicated and amplified to create the change the community needs.

1. U.S. Food and Drug Administration: <https://www.fda.gov/drugs/development-approval-process-drugs/fda-patient-focused-drug-development-guidance-series-enhancing-incorporation-patients-voice-medical>. Accessed August 2023

2. Guide to Patient Involvement in Rare Disease Therapy Development; A Publication of the Rare Disease PFDD Compendium Workshop Series, The Everylife Foundation for Rare Diseases, 2022.