#708LPB

Clinical Trial Readiness in a Rare and Underserved Disease: Learnings from Community Engagement and the Lived Experience in Becker Muscular Dystrophy (Becker)

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Background

Becker muscular dystrophy (Becker) is a serious, irreversible, and potentially fatal neuromuscular disorder with high unmet medical need[1]. There are no approved therapies for Becker and only two interventional trials are actively recruiting, highlighting the limited research exclusively dedicated to this indication. There are no Becker-specific advocacy organizations to facilitate clinical trial readiness, promote research or serve as a patient-focused resource for clinical trial education and awareness. This has led to a fragmented and disenfranchised community, making research and trial recruitment challenging.

Objectives

This patient-centered survey aimed to understand the lived experience and patient journey of those living with Becker as a foundation for developing strategies to educate and activate the Becker community.

Approach

A survey, developed with feedback from the patient community, was administered by a third-party health research agency, Engage Health, to ensure blinding.

The survey was reviewed by a Becker Patient Council supported by Edgewise Therapeutics, to ensure comprehension and ease of completion.

Participants were recruited by Engage Health using a variety of methods including social media platforms, Becker educational events, and individual promotion and outreach.

Survey design, conduct and analysis reflects best practices as outlined in the Food and Drug Administration guidance for incorporating the patient voice into drug development.[2,3]

Outcomes

Participant Characteristics (n=50)

- Individuals living with Becker (n=23, 46%)
- Primary caregivers (n=11, 22%)
- Dyads (n=5, 10%)
- Participants represented 22 US states.

Ambulation status varied across ages, and more mobility support was needed over the course of the disease

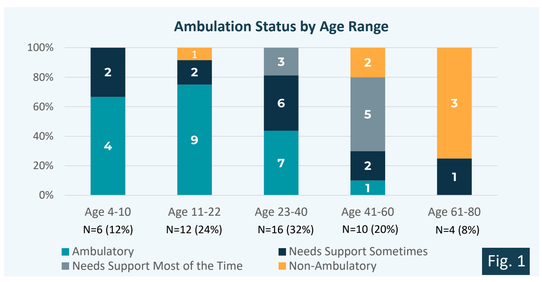

Fig. 1

Misdiagnosis and delays in diagnosis were common

- 36% were initially misdiagnosed.
- 58% lived with symptoms for over a year before finding a diagnosis.
- 16% did not receive a diagnosis for over ten years.
- Having an immediate family member with a Becker diagnosis improved time to diagnosis.
- Those with no family member with Becker (n=23) had a heterogenous experience with receiving a diagnosis, ranging from 1 to 10 years.

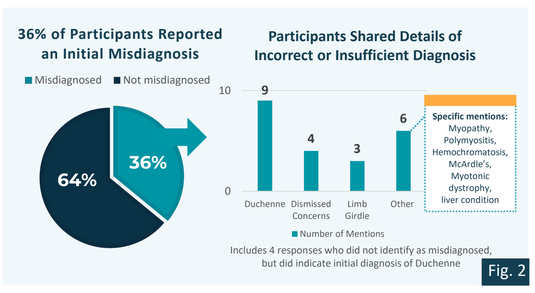

Fig. 2

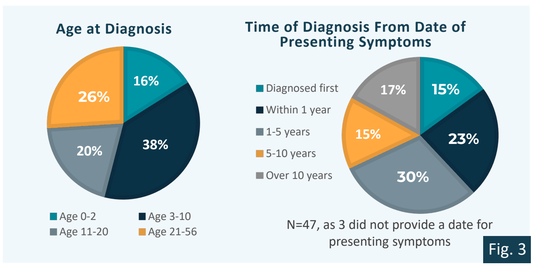

Fig. 3

- *"With an earlier diagnosis, I could have avoided school and job activities that taxed my muscles like running or lifting heavy objects."*

-Individual living with Becker

The most common symptoms related to mobility were difficulty with walking and rising from a chair

- Mobility devices were commonly used to support participants experiencing these symptoms.
- Walking sticks/canes and powerchairs were the most used mobility aids.
- Of those 16 years and older, 70% reported using a mobility device at least part time.

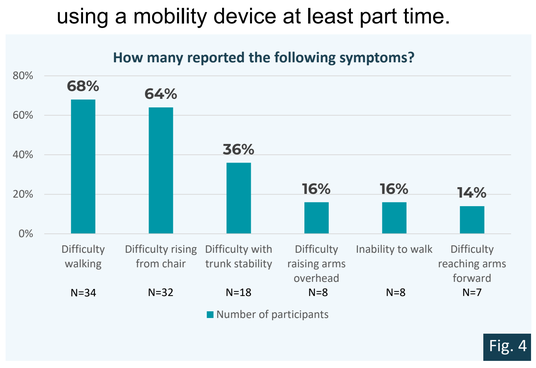

Fig. 4

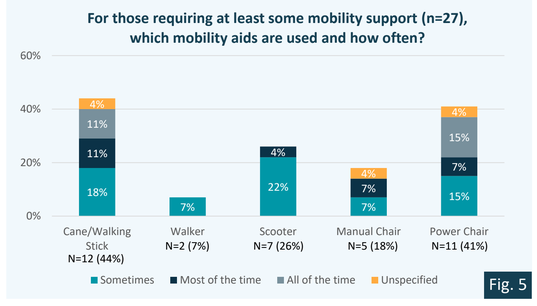

Fig. 5

- Many physical symptoms, including difficulty walking, rising from chair, difficulty with trunk stability, became more prevalent in early adulthood (between ages 23 and 40), when consistent specialist care was seen to drop off.
- *"I have been told I am not bad enough yet and there are not enough slots at the clinic"*

-Individual living with Becker

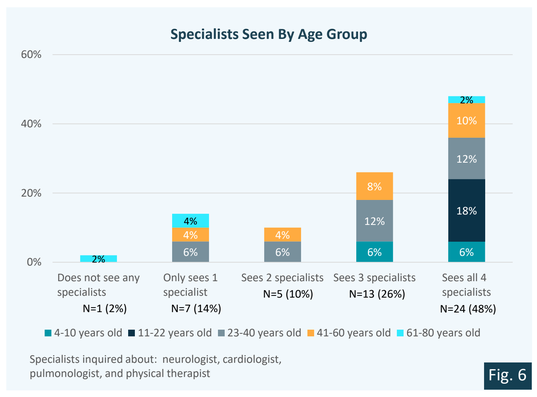

Fig. 6

Cardiac testing was inconsistent despite heart health being the most pressing concern noted by participants

- 78% saw a cardiologist as part of routine care.
- Those who reported only seeing a cardiologist "as needed" or who did not see a cardiologist had more limited mobility.
- 98% of respondents reported receiving cardiac testing, however the types and frequency of testing varied greatly.
- No respondents over age 38 received CMRI/MRI.
- *"My heart health is what actually threatens my health."*

-Individual living with Becker

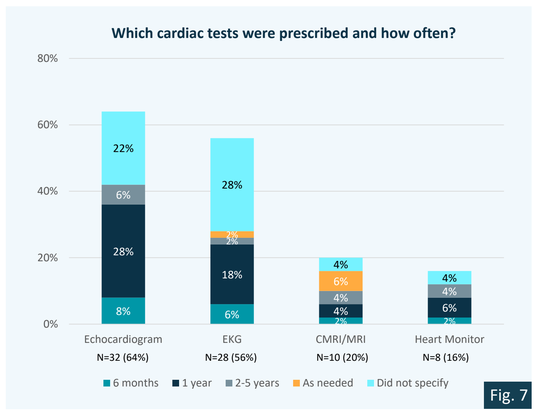

Fig. 7

Majority reported experiencing pain and fatigue

- Management for these are primarily self-driven without input from a healthcare provider.
- Most common pain management approach referenced was over the counter medication.
- 22% reported the pain they experience never fully goes away, and 42% noted that when it occurs it will last for several hours.
- *"All day I am struggling to perform daily activities, and I deal with fatigue all day."*

-Individual living with Becker

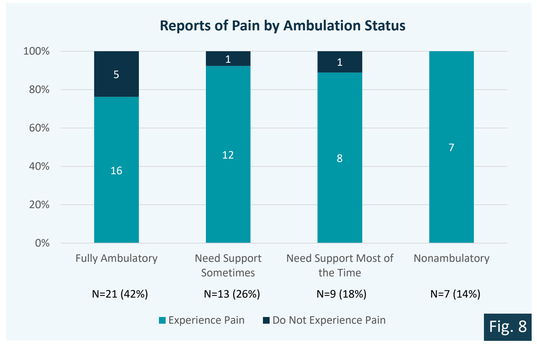

Fig. 8

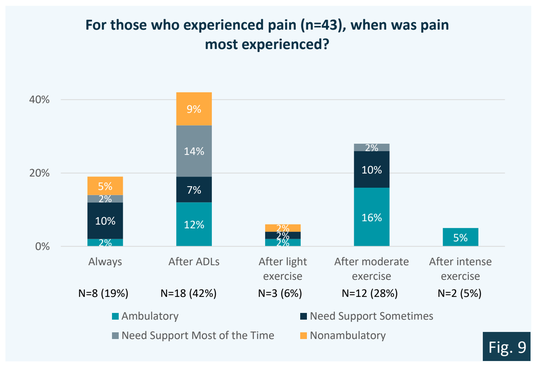

Fig. 9

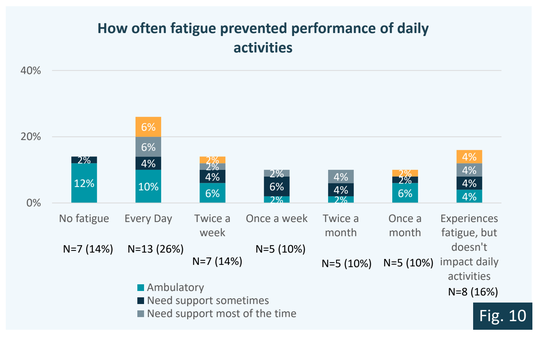

Fig. 10

Addressing physical symptoms and advancing research were the most common themes for improving quality of life; need for psychosocial support was also commonly mentioned

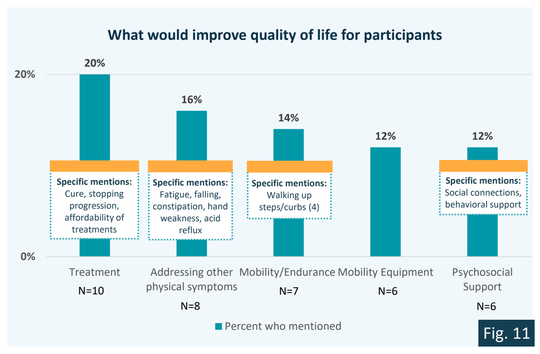

Fig. 11

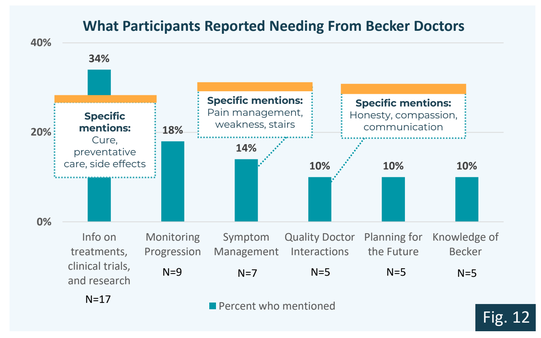

Fig. 12

Discussion

- The outcomes of this community engagement demonstrate the underserved nature and lack of sufficient resources in the Becker community.
- Inconsistent care and symptom management has physical and emotional impacts on individuals living with Becker.
- Individuals living with Becker desire education so that they can be active participants in their care and advocate for their needs.
- Data from this study provided a framework for educational efforts for patients and clinicians, led to collaborations with advocacy organizations, and inspired new community initiatives.
- An informal collaboration of stakeholders organized the Becker Education and Engagement Day, first occurring in 2023 at multiple locations across the US, which addressed many of the information gaps noted in this survey.
- In Europe, TREAT-NMD has taken the initiative to bring this event to two locations in Europe in 2024.
- This survey has highlighted the need for the Becker community to be more informed and proactive about their disease, best care practices, and how their voices as a community can accelerate research and move the clinical trial ecosystem forward.

Acknowledgements
The authors are grateful to the participants in this study.

References
[1] Andrews, J.G.D., M. F. Meaney, F. J., Correlates of care for young men with Duchenne and Becker muscular dystrophy. Muscle and Nerve, 2014. 49(1): p. 21-25
[2] 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 September, 2024.;
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