

# Burden of Duchenne Muscular Dystrophy in Patients with Mutations Amenable to Exon 44 Skipping (DMD44)



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## Study Objectives

- Identify patient-centered outcomes.
- Document the burden of disease for patients with DMD44.
- Evaluate time from symptom onset to diagnosis and attributes important to quality of life.

## Study Design

- 10 individuals with DMD44 and caregivers were recruited to participate.
  - Five ambulatory and five non-ambulatory.
- Survey sent to participants prior to focus group.

## Methods

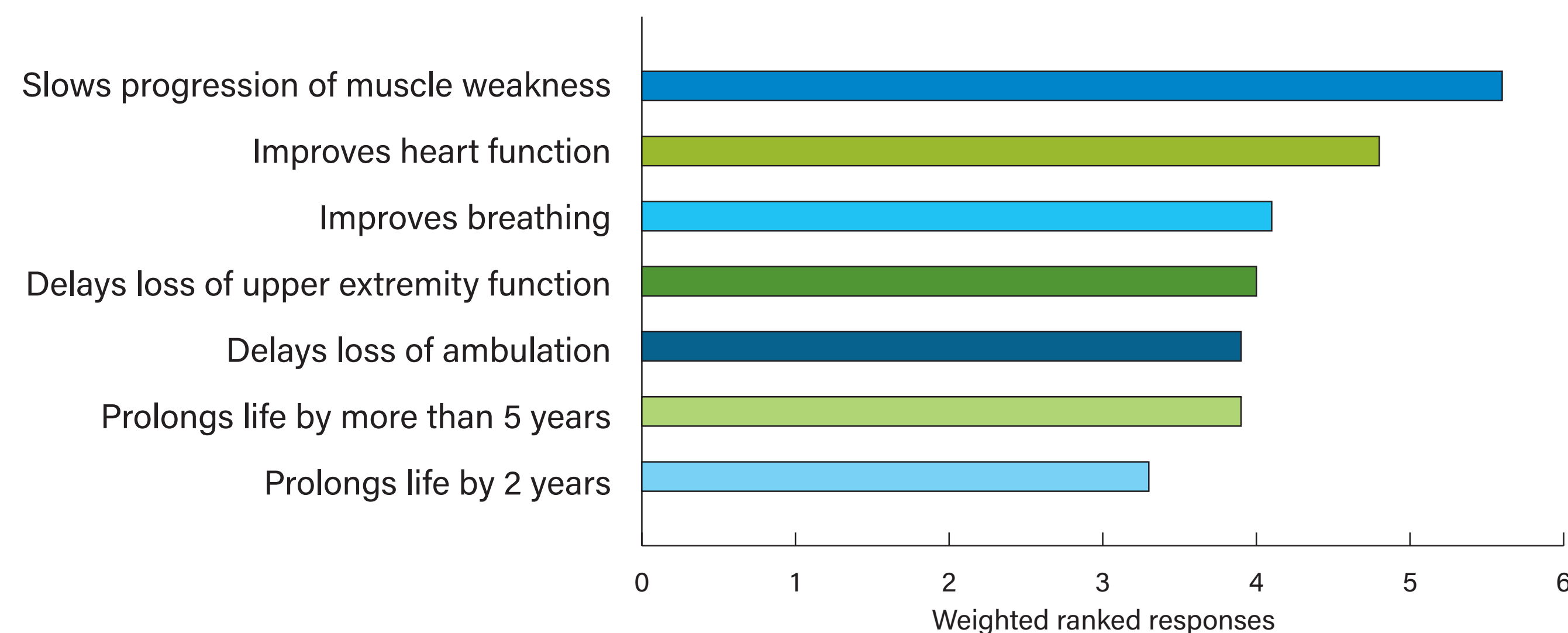
- Survey examining the following issues:
  - Burden of DMD44.
  - Preferred treatment attributes.
  - Patient-centered clinical endpoints.
  - Preferred approaches to measure stabilization of disease.
  - Time from symptom presentation to diagnosis.

## Results

### Demographics

Group	Age (time of study)	Age at time of diagnosis
Ambulatory	9, 10, 10, 15, 19	2.5, 5, 5, 6, 7
Non-Ambulatory	14, 18, 19, 20, 31	1, 2.5, 4, 6, 7

### Most Desirable Attributes for "New" Treatments



### Attributes Important to Quality of Life (Abridged Quotes by Patients and Caregivers)

#### Ambulatory

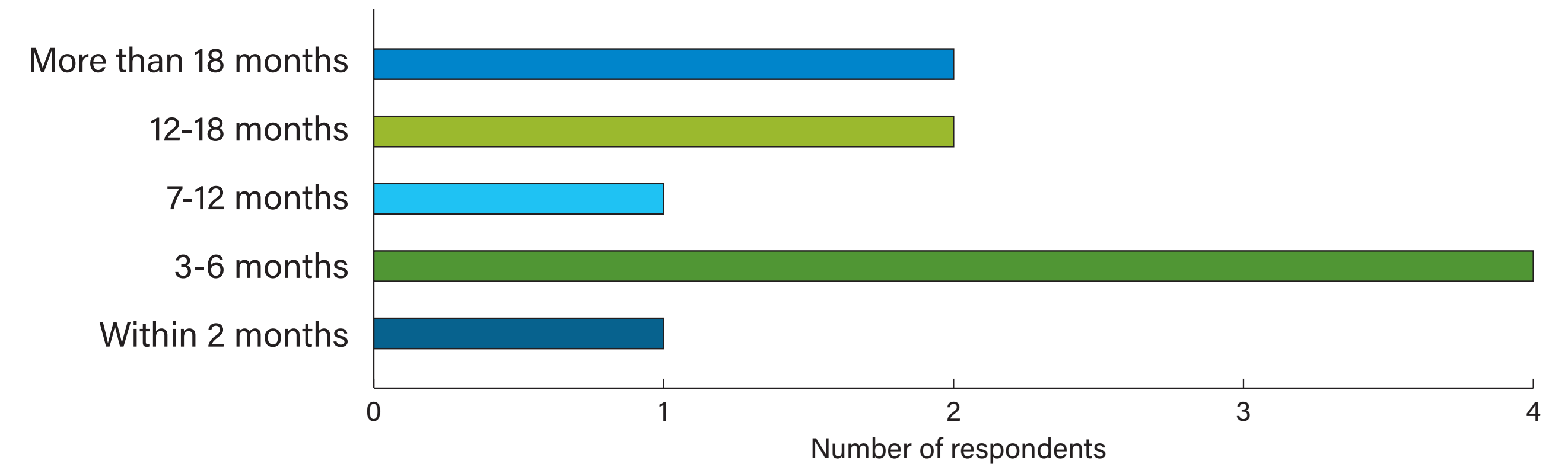
- Finding ways to see new things, go new places, try new things.
- Being able to play with his friends and brothers.
- Staying ambulatory, overall healthy body function.
- Safety, inclusion, security, acceptance.

#### Non-Ambulatory

- Interacting in virtual worlds to replace relationships in the physical world. We hear him laughing and being challenged in his online games and that makes us happy he has found happiness.
- The more things I can do for myself the better. Eating, brushing teeth, moving around in bed.
- Cardiac and respiratory stability and being able to feed myself.

## Results (continued)

### Time from Symptom Onset to Diagnosis



### Attributes Important to Patients and Caregivers to Assess Disease Stabilization (Abridged Quotes by Patients and Caregivers)

#### Ambulatory

- Not progressing to a wheelchair. Keep walking, running, taking stairs!
- Keep walking the same amount. Lifting same amount of weights. Validating pulmonary or heart functions have not decreased.
- Timed tests to see how they do running, rising from the stairs, standing from sitting.

#### Non-Ambulatory

- At least delay loss of ambulation, heart failure, lung function.
- Maintaining your current skills i.e., lifting a cup, brushing teeth, combing your hair, give a hug.
- Ability to self-feed, stand or sit for bathing and toilet activity.
- Echocardiogram, DEXA scan, respiratory evaluation.
- Eat without help, no loss of upper extremity muscles strength, stable pulmonary function tests.

### Important Outcomes for DMD44 Provided by Patients and Caregivers

#### Ambulatory

- Grip strength
- Use of joystick and keyboard
- Upper body strength
- Lung function/breathing ability
- Cardiac function
- Leg lift and climbing stairs
- Standing up from the floor
- Number of falls

#### Non-Ambulatory

- Use of joystick and keyboard
- Ability to feed myself
- Ability to drive motorized wheelchair
- Ability to scratch head
- Lung function/breathing ability
- Heart function
- Lifting arms

## Conclusions

- Disease stabilization more important than symptom improvement.
- Burden of disease varies by ambulatory status.
- Significant differences between ambulatory and non-ambulatory.
- The odyssey from symptom to diagnosis can be lengthy.
- Physical burdens impact quality of life of patients and caregivers.
- DMD44 affects patients and caregivers emotionally and socially.
- Virtual games allow patients to maintain social relationships.

