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

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.