Understanding Becker Muscular Dystrophy: Disease Overview & Natural History

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Disease overview[1-3]

Becker muscular dystrophy (Becker)

Is a serious, rare, neuromuscular disease that can result in irreversible loss of muscle function and can lead to loss of ambulation.

Becker is an X-linked, recessive disorder caused by a gene mutation impacting dystrophin and driven by contraction- induced muscle damage. As muscle damage accumulates, it can be more difficult to perform everyday activities like walking or climbing stairs.

Becker is a multi-system disease that can impact more than just ambulation. For example, patients may develop cardiovascular complications as well.

Muscle loss and functional decline

Can begin at any age in Becker patients. Once decline begins, it continues, so early diagnosis and disease management is crucial to help preserve muscle and overall function.

There are currently no approved treatment options for Becker.

Tracking the natural history of Becker

The NSAA is a functional assessment that measures an individual's ability to perform 17 items that have real-world implications for individuals with Becker.[3-6]

Real-world implications for individuals with Becker

Measure	Activity
Jump, hop, run	Playing sports
Stand on heels	Walking on uneven ground, cycling, difficulty getting out of a chair, striding, cycling
Rise from floor	Getting up after falling, playing on the floor with children
Climb box steps	Independent outdoor mobility particularly easy tasks like stairs and sidewalk curbs
Stand on one leg	Dressing oneself, putting on shoes/socks while standing, reaching high shelves
Gets to sitting	Sitting up in bed, adjust to falls
Rise from chair	Using a toilet independently, getting out of bed, using public transportation to get around
Walk	Walking to mailbox to pick up mail, hiking, everyday mobility
Stand	Grooming, preparing meals, adapting to mobility device, transferring to chair

The NSAA

The NSAA consists of 17 items to evaluate motor function.

- Hop left leg
- Hop right leg
- Stand on heels
- Rise from floor
- Run
- Jump
- Lift head
- Descend box step (L)
- Descend box step (R)
- Climb box step (L)
- Climb box step (R)
- Stand one leg (L)
- Stand one leg (R)
- Get to sitting
- Rise from chair
- Walk
- Stand

Each activity is scored on whether it can be completed

+2 pts	Perform normally
+1 pts	Perform with compensation due to weakness
0 pts	Cannot perform

The NSAA is a clinically meaningful measure of function

For individuals living with Becker, a 1-point change in NSAA can mean the loss of independence in one or more activities of daily living. For example:

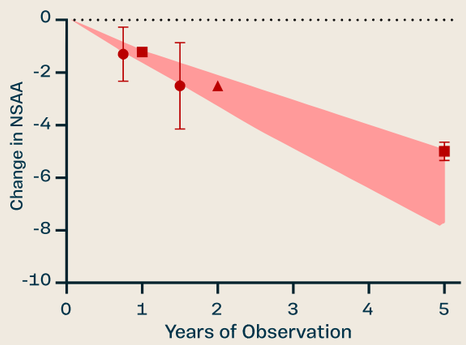
From using stairs or steps to requiring assistance from another person or mobility device.

From using the toilet independently to asking for help to get up from the toilet.

From being able to get up from a fall to requiring someone else's help to get back up.

The NSAA is utilized in muscular dystrophy natural history studies to longitudinally assess function and disease progression.[3-6]

Persistent decline in Becker muscular dystrophy



- De Wel B, et al. (ambulatory)[4]
- Bello L, et al. (ambulatory, NSAA 10-32)[6]
- Van de Velde NM. et al. (ambulatory)[5]

95% CI shown

Studies demonstrate an average decline in NSAA score of

0.9 to 1.7

points per year.[3-6]

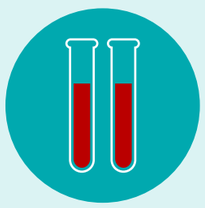
Slowing or stabilisation of disease progression is a key target for Becker research.

Diagnosing Becker

Because Becker is a progressive, irreversible disease, early diagnosis and disease management is important to help preserve muscle and overall function.

Diagnosis of Becker can include:

Patient assessment of weakness, functional tests, family history of Duchenne or Becker



Lab tests showing elevated biomarkers of muscle injury, such as creatine kinase

Genetic testing with the identification of a pathogenic variant in the *DMD* (*dystrophin*) gene

Becker often undiagnosed, misdiagnosed, or diagnosed late

Awareness about Becker in the general healthcare community remains a challenge and Becker is often mistaken for Duchenne muscular dystrophy (Duchenne) or referred to as a milder form of Duchenne. However, Becker is not Duchenne.

Investigational agents are currently in clinical development for the treatment of Becker.

Key Points

Becker is a serious, rare, progressive neuromuscular disease that leads to loss of muscle and impaired motor function.

The NSAA is an assessment used in natural history studies to longitudinally assess disease progression and functional decline in Becker. Current studies demonstrate that individuals with Becker experience an average decline of 0.9–1.7 NSAA points per year.

The NSAA is a clinically meaningful assessment of function. Each NSAA item has real-world implications for Becker patients and score changes can represent significant impacts to a patient's ability to perform activities of daily living.

Research shows that once decline begins in individuals with Becker, it is continuous and irreversible, so early diagnosis and disease management is crucial to help preserve muscle and overall function.

Abbreviations:

Becker: Becker muscular dystrophy; L: left; NSAA: North Star Ambulatory Assessment; Pts: points; R: right.

References:

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