NxGen MDx

DMD-Related Dystrophinopathy (Duchenne Muscular Dystrophy and Becker Muscular Dystrophy)

What Your Results Mean

Test results indicate that you are a carrier of a *DMD*-related dystrophinopathy (Duchenne muscular dystrophy and Becker muscular dystrophy). Females carriers are usually not affected with Duchenne or Becker muscular dystrophies because they make enough of the dystrophin protein. However, female carriers can have some symptoms of Duchenne, such as changes to heart function, mild muscle weakness, fatigue, or cramping in their muscles. Female carriers have an increased chance to have a child with a *DMD*-related dystrophinopathy. Consultation with a genetic counselor for a more detailed risk assessment is recommended.

We recommend that you share this information with all of your health care providers. Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning and their own personal clinical management.

Recommended Next Steps

Male children have a 50% risk to have a *DMD*-related dystrophinopathy and female children have a 50% risk to be carriers for a *DMD*-related dystrophinopathy. In the absence of symptoms, reflexive testing of your male partner or donor for *DMD*-related dystrophinopathies is not indicated due to the X-linked inheritance pattern of the condition.

DMD-Related Dystrophinopathy (Duchenne Muscular Dystrophy and Becker Muscular Dystrophy) Explained

What is a *DMD*-Related Dystrophinopathy?

Duchenne Muscular Dystrophy

Duchenne muscular dystrophy (DMD) is typically diagnosed in early childhood, with first symptoms being identified as general motor delays, gait problems, delays in walking, and learning difficulties. Due to the progressive nature of DMD, children with DMD are usually wheelchair dependent by 12 years of age. Nearly all individuals with DMD are diagnosed with cardiomyopathy by age 18. Cognitive impairment characterized by low IQ and difficulty with short term verbal memory is also common. Most individuals with this condition do not survive past the age of 30.

Becker Muscular Dystrophy

Becker muscular dystrophy (BMD) is characterized by mild and later-onset symptoms in comparison to DMD. Symptoms of muscle weakness typically occur much later than in DMD; some individuals with BMD may not experience mobility issues until their late twenties. While muscle weakness is milder in individuals with BMD in comparison to DMD, cardiomyopathy is still a concern. Heart failure is the most common cause of death in individuals with BMD. Most individuals with this condition survive to their mid-forties.

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Prognosis

The prognosis for Duchenne muscular dystrophy is variable, but most males will be wheelchair-dependent by age 13 and die before 30 years of age due to heart or respiratory failure. Males who have Becker muscular dystrophy have a longer life expectancy reaching into their forties or fifties. Prognosis in females is generally better, but lifespan may still be shortened in the presence of dilated cardiomyopathy.

Treatment

Management of the disease is generally focused on a combination of physical therapy, medication, and regular cardiac and respiratory screenings. Female carriers should be treated by a cardiologist.

Resources
Muscular Dystrophy Association
https://www.mda.org/
Genetics Home Reference
https://ghr.nlm.nih.gov/condition/duchenne-and-becker-muscular-dystrophy
National Society of Genetic Counselors
https://www.nsgc.org/

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