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My46 Trait Profile

Duchenne / Becker Muscular Dystrophy

Other Names: Dystrophinopathy

Duchenne muscular dystrophy and Becker muscular dystrophy are genetic conditions characterized by progressive muscle disease. They are both caused by mutations in the DMD gene which makes the dystrophin protein.

Characteristics of Duchenne / Becker Muscular Dystrophy

Duchenne Muscular Dystrophy (DMD) and Becker Muscular Dystrophy (BMD) are both types of muscle disease caused by changes or mutations in the DMD gene. DMD has an earlier onset than BMD; early signs usually include boys ages 3-5 years with delays in meeting developmental milestones. Boys may exhibit delays in walking and speech, have frequent falling, begin walking on tiptoes, and/or have learning difficulty as compared to other children of the same age. Frequently, boys with DMD use their hands to help them get up from a seated position on the floor (Gower's maneuver) due to weak muscles in their hips. Calf muscles frequently appear large (calf pseudohypertrophy), but this is not due to extra muscle growth. Boys with DMD have continued muscle breakdown over time usually needing to use a wheelchair full time by age 12. Since the heart is also a muscle, boys with DMD are also at risk for dilated cardiomyopathy (thinning of the heart walls), as well as electrical changes of the heart. Boys with DMD also may have learning difficulties and/or autism spectrum disorder.

BMD has a similar but milder signs and symptoms and a slower progression of muscle breakdown than seen in boys with DMD. Boys with BMD can be diagnosed at any time from childhood to adulthood. Boys with BMD are also at risk for heart symptoms like those seen in DMD. Learning difficulties are not found as often in boys with BMD.

Diagnosis/Testing

Initial testing in a boy suspected to have DMD or BMD often involves measuring an enzyme specifically found in muscles, Creatine Kinase (CK) or Creatine Phosphokinase (CPK). This enzyme is released in to the blood stream when muscles break down. Therefore, boys with both DMD and BMD have elevated levels of CK in the blood. Genetic testing can also be done to try to find any changes or mutations in the DMD gene. The DMD gene makes a protein called dystrophin. Depending on where these mutations are found in the DMD gene, they are passed on to the protein dystrophin. Some mutations can cause the body not to make any dystrophin protein. These are changes more frequently seen in boys with DMD. Other changes can cause the body to make less dystrophin or a smaller dystrophin protein that may or may not work properly in muscle cells. These changes are more often seen in boys with BMD.

Although most boys with DMD/BMD have identifiable DMD gene mutations, if a mutation is not found, a muscle biopsy may be done to measure the amount of the protein dystrophin in the muscle cells. A muscle biopsy can also be used to help differentiate DMD vs. BMD by measuring the amount and size of the dystrophin protein in the muscle of affected boys.

Management/Surveillance

The management of a family with DMD/BMD involves many specialties including neurology, physical/occupational therapy, orthopedics, cardiology, pulmonology, and nutrition. Certain medications (i.e., corticosteroids) are often recommended to help maintain walking and may even be continued after boys become solely

dependent on using a wheelchair in order to help maintain upper body strength. It is recommended that boys with DMD and BMD be followed closely to monitor any orthopedic problems, cardiac symptoms, scoliosis, breathing problems, and nutrition in order to help maintain a healthy weight. Great care is given to maintain independence and quality of life for boys with DMD and BMD.

Boys with DMD and BMD can be at risk for increased muscle break down (rhabdomyolysis) with excessive exercise. Boys should watch for urine the color of cola (myoglobinuria). If this happens, boys should be taken to the emergency room for hydration to avoid kidney damage and/or sudden death. It is frequently recommended that anyone with a muscle disease avoid anesthetic agents due to the risk of malignant hyperthermia (dangerous increase in body temperature).

Mode of inheritance

DMD/BMD is inherited in an X-linked recessive pattern. This means the DMD gene is located on the X chromosome. Females have two X chromosomes, and males have one X chromosome and one Y chromosome. Males with a change or mutation in the DMD gene will be affected with muscle disease, while females with a mutation in the DMD gene on only one copy of their two X chromosomes are considered carriers of DMD/BMD. Some carrier females (approximately 20-30%) may have mild symptoms of DMD/BMD. The mutation can be inherited from a carrier mother or it can occur brand new (de novo) in an affected child.

Risk to family members

The risk for a carrier mother to pass on her DMD gene mutation to her children is 50% with every pregnancy. If that child is male, he will be affected with muscle disease; while if that child is female, she will be a carrier. A non-carrier mother has a residual risk of 15% to have another child with DMD/BMD due to a phenomenon called gonadal mosaicism. Gonadal mosaicism occurs when a DMD gene mutation is found in a mother's egg cells, but not all of the cells of the body.

Special considerations

Although most females with mutations in the DMD gene are considered carriers and do not have symptoms, some (about 20-30%) may have muscle or heart symptoms within their lifetime. It is recommended that beginning at age 25, women that are carriers of DMD see a cardiologist every few years to have their heart examined.

Resources

Centers for Disease Control and Prevention: Muscular Dystrophy
http://www.cdc.gov/ncbddd/musculardystrophy/index.html
Muscular Dystrophy Association (MDA)
http://www.mdausa.org
DuchenneConnect Patient Registry
http://www.duchenneconnect.org
Parent Project Muscular Dystrophy
http://www.parentprojectmd.org

References

Bushby, K. et al. (2010a)."Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and pharmacological and psychosocial management." Lancet Neurology 9(1): 77-93.
Bushby, K. et al. (2010b)."Diagnosis and management of Duchenne muscular dystrophy, part 2: implementation of multidisciplinary care." Lancet Neurology 9(2): 177-189.
Darras BT, Miller DT, Urion DK. (Updated 23 November 2011). Dystrophinopathies. In: GeneReviews at GeneTests Medical Genetics Information Resource (database online). Copyright, University of Washington, Seattle. 1997-2013. Available at http://www.ncbi.nlm.nih.gov/books/NBK1119/. Accessed [04/02/2013].
Kang PB, Kunkel LM. The muscular dystrophies. In: Scriver CR, Beaudet AL, Sly WS, Valle D, Vogelstein B, eds. The Metabolic and Molecular Bases of Inherited Disease (OMMBID). New York: McGraw-Hill; Chap 216. Print.

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