

Duchenne (Pseudohypertrophic) Muscular Dystrophy

DMD is the most severe and the most common MD of childhood. It is inherited as an X-linked recessive trait, and the single-gene defect is located on the short arm of the X chromosome. DMD has a high mutation rate, with a positive family history in about 65% of cases. Genetic counseling is an important aspect of the care of the family. In about 30% of cases, it is a new mutation, and the mother is *not* the carrier ([Sarnat, 2016b](#)).

As in all X-linked disorders, males are affected almost exclusively. The female carrier may have an elevated serum creatine kinase, but muscle weakness is usually not a problem; however, about 10% of female carriers develop cardiomyopathy ([Manzur, Kinali, and Muntoni, 2008](#)). In rare instances, a female may be identified with DMD disease yet with muscular weakness that is milder than in boys ([Sarnat, 2016b](#)). At the genetic level, both DMD and Becker MD (a milder variant) result from mutations of the gene that encodes dystrophin, a protein product in skeletal muscle. Dystrophin is absent from the muscles of children with DMD and is reduced or abnormal in children with Becker MD. Children with Becker MD have a later onset of symptoms, which are usually not as severe as those seen in DMD. The incidence is approximately 1 in 3600 male births for the Duchenne form and approximately 1 in 30,000 live births for the Becker type ([Sarnat, 2016b](#)). [Box 30-8](#) describes the characteristics of DMD.

Box 30-8

Characteristics of Duchenne Muscular Dystrophy

- Early onset, usually between 3 and 7 years old
- Progressive muscular weakness, wasting, and contractures
- Calf muscle pseudohypertrophy in most patients
- Loss of independent ambulation by 9 to 12 years old