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Editorial

Dystrophinopathy

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EDITORIAL

Duchenne muscular dystrophy, Becker muscular dystrophy, and DMD-associated dilated cardiomyopathy are among the dystrophinopathies, which span a range of X-linked muscle illness ranging from moderate to severe (DCM). The phenotypes of asymptomatic rise in serum creatine phosphokinase (CK) and muscular cramps with myoglobinuria are at the mild end of the range. Progressive muscle disorders are categorised as Duchenne/Becker muscular dystrophy when skeletal muscle is predominantly damaged and as DMD-associated dilated cardiomyopathy (DCM) when the heart is primarily afflicted at the severe end of the range.

Early-onset Duchenne muscular dystrophy (DMD) is characterised by delays in motor milestones such as walking independently and rising up from a supine position. A waddling stride and difficulties ascending stairs, sprinting, jumping, and rising up from a crouching position are all symptoms of proximal weakness. DMD progresses quickly, and by the age of 12, afflicted children are wheelchair-bound. After the age of 18, approximately everyone with DMD develops cardiomyopathy. Respiratory problems and worsening cardiomyopathy are frequent causes of mortality in people in their third decade. Becker muscular dystrophy (BMD) is characterised by skeletal muscle weakening that develops later in life. Men who develop symptoms after the age of 30 years and remain ambulatory well into their 60s have been identified as being on the moderate end of the spectrum, thanks to improved diagnostic procedures.

Heart failure from DCM is a frequent cause of morbidity and the most prevalent cause of mortality in BMD, despite the lesser skeletal muscle involvement. The average age of death is in the mid-forties.

DIAGNOSIS

Identification of a hemizygous pathogenic variant in DMD on molecular genetic testing in a male and of a heterozygous pathogenic variant in DMD on molecular genetic testing in a female establishes the diagnosis of a dystrophinopathy in a proband with the characteristic clinical findings and elevated CK concentration and/or identification of a heterozygous pathogenic variant in DMD on molecular genetic testing in a female. Females might have a typical dystrophinopathy or be asymptomatic carriers of the gene.

Treatment Of Manifestations

In both the DMD and BMD phenotypes, ACE inhibitors are used with or without beta blockers to treat cardiomyopathy. Diuretics and oxygen are used to treat congestive heart failure, and cardiac transplantation is an option for those with severe dilated cardiomyopathy and BMD who have little or no clinical signs of skeletal muscle degeneration. Bracing and surgery are used to treat scoliosis. Individuals with DMD between the ages of five and fifteen years benefit from corticosteroid medication; the same treatment is used in BMD, albeit the efficacy is less evident.

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