
DETECTING SIGNS AND SYMPTOMS OF DUCHENNE MUSCULAR DYSTROPHY

For many families, getting a Duchenne muscular dystrophy (DMD) diagnosis involves a journey. The diagnosis may come as a result of genetic testing in a family that reveals an inheritance pattern, or it may come after seeking a cause for certain signs or symptoms in a child. Knowing what DMD is and how to detect it can help speed up the process of getting a diagnosis.

What is Duchenne Muscular Dystrophy?

DMD is an inherited genetic disease, meaning it is caused by a variant (mutation) in a gene that is passed from a parent to a child. The gene variant results in a missing protein, called dystrophin, in muscle cells. When a person doesn't have enough dystrophin protein, their muscles get progressively weaker and leak the enzyme creatine kinase (CK) into the blood. An elevated CK level is a sign of DMD.

DMD affects 1 out of every 5,000 male births. Children with DMD — nearly always boys — may not show obvious signs of the disease in infancy; symptoms typically become apparent around age 3. For more information on DMD, check out the DMD Disease Fact Sheet in English or Spanish.

Prenatal Testing

There are no specific signs or symptoms related to DMD during pregnancy, although prenatal testing may detect whether a baby has the disease. Your obstetrician or genetic counselor may recommend prenatal testing if there is a history of DMD in your family.

Testing in Infants

There are no visible signs of DMD in infants. Babies with DMD have elevated CK levels, but muscle enzyme tests are not routinely done by pediatricians.

Newborn screening is a public health program that screens babies when they are born for certain diseases detectable at birth for which there are treatments that could prevent harmful health impacts. Currently, the technologies for newborn screening for DMD are under development and hopefully will be ready soon for widespread adoption. Once ready, MDA will advocate to add DMD to the Recommended Uniform Screening Panel (RUSP), which is the national guideline for newborn screening programs.

Until newborn screening is available, parents who know DMD runs in their family should talk with their pediatrician about a muscle enzyme test for their baby. **Studies show that treatments for DMD may be most effective when administered early in the disease process.**





Signs and Symptoms

Children with DMD usually reach early milestones on time or slightly delayed. For instance, infants with DMD can raise their heads, though parents may notice a delay in their ability to roll over and crawl. A delay in learning to walk also is common with DMD, as are walking on toes and constipation.

By age 3, difficulty walking, running, and jumping usually become apparent. These often are the first noticeable signs of DMD, along with enlarged calves and a waddling gait.

Care for DMD

If you suspect your child has DMD, ask your pediatrician to check his muscle enzymes. If the test shows a high CK level, consult a pediatric neurologist for a possible DMD diagnosis and to discuss care options. Your pediatrician can refer you to a pediatric neurologist, or you can find one through an MDA Care Center. (Search for MDA Care Centers at mda.org/carecenters.)

It's best to seek testing and care early if you have a family history of DMD or suspect any developmental delays in your child.

To test for DMD in infants, a blood sample is collected with a heel stick.

To learn more about DMD, visit mda.org or contact the MDA Resource Center at 833-ASKMDA1.

