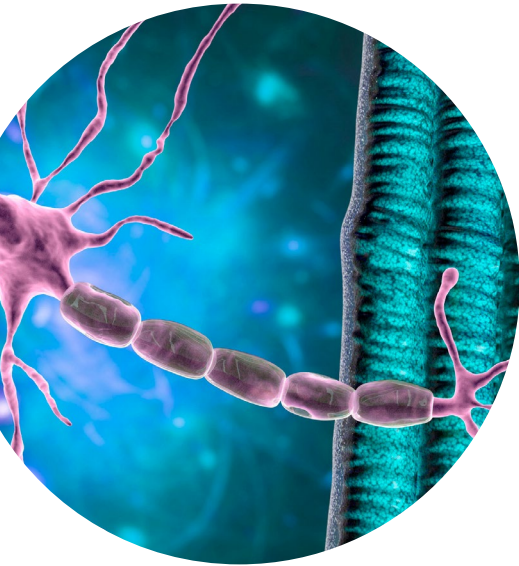


How Duchenne Muscular Dystrophy Happens

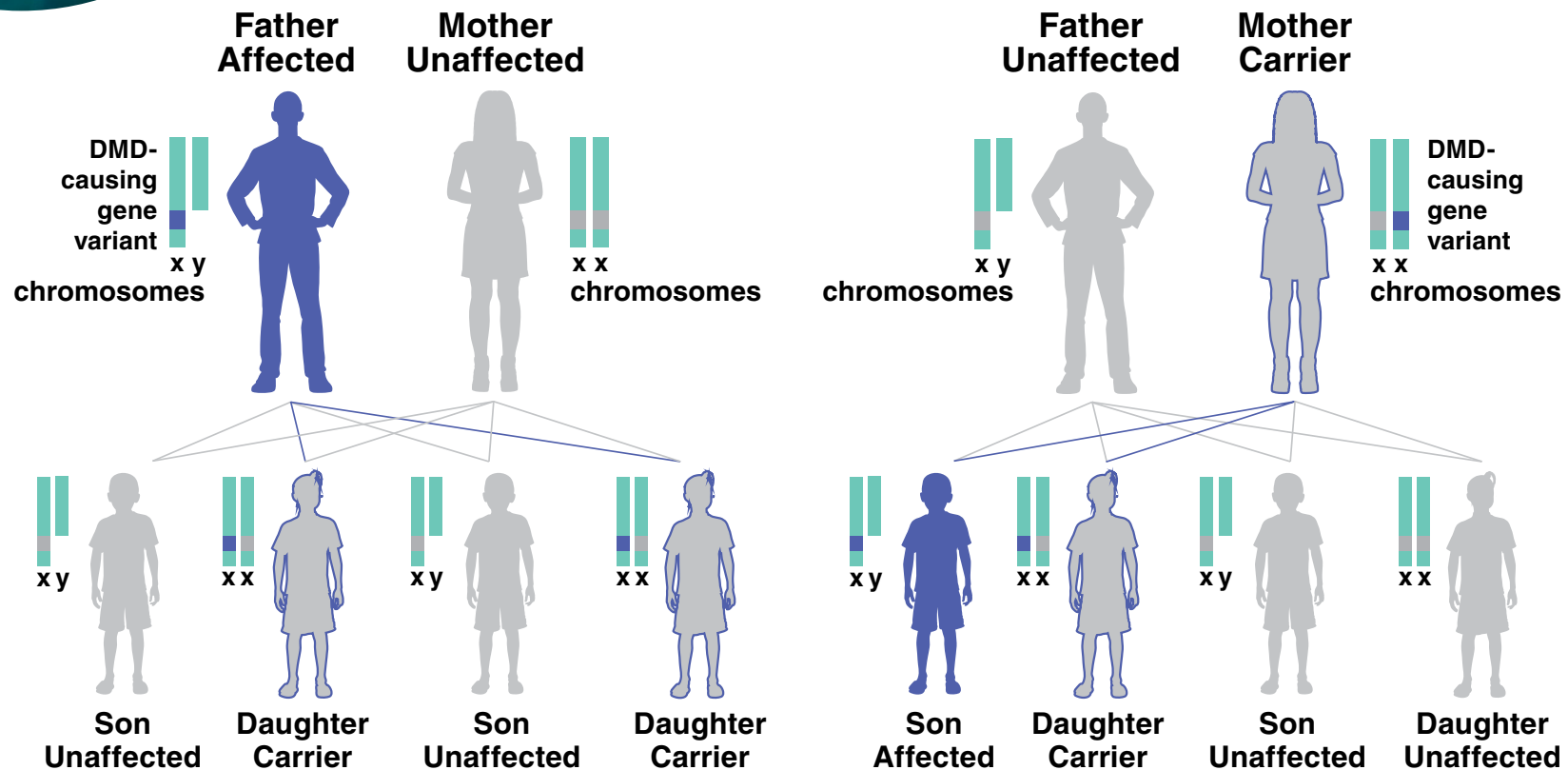


We have thousands of genes in our bodies that help with different functions. Duchenne muscular dystrophy (DMD) is caused by a change, called a gene variant, in the dystrophin (*DMD*) gene. This gene encodes a protein, also called dystrophin, which plays an important role in the structure and strength of skeletal and heart muscles. A variant in the *DMD* gene can result in dystrophin protein that doesn't work correctly, affecting the muscles.

Inheritance pattern

The *DMD* gene is located on the X chromosome. Because males have only one copy of the X chromosome, boys who inherit the disease-causing gene variant will have DMD. Females who inherit the gene variant on one of their X chromosomes will be carriers.

About one-third of individuals with DMD have no family history, meaning both mother and father are unaffected. This is called de novo mutation.



Genetic Testing for DMD

What does genetic testing do?

Many neuromuscular diseases have similar symptoms. Testing can confirm a diagnosis of DMD and determine the type of genetic variant causing it.

Different types of *DMD* gene variants may impact the protein to a lesser or greater extent, which leads to a range of physical presentations in the dystrophinopathies (conditions caused by variants in the *DMD* gene).

Knowing your specific diagnosis is important as it allows you to understand potential health implications.

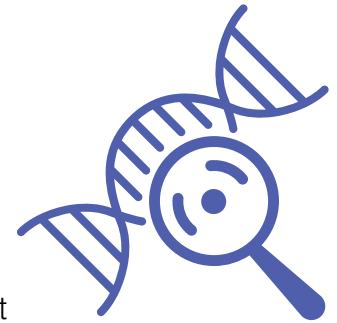
It also allows your doctor to know which therapies might be most suitable for your specific variant. Lastly, knowing your diagnosis can empower you to seek out resources and clinical trials that could be relevant for you.



Why do I need a genetic test?

A genetically confirmed diagnosis is important because:

- ▶ Genetic testing can determine the type of DMD and provide other information that helps doctors understand what the course of the disease is likely to be.
- ▶ Knowing your gene variant is important as it can help doctors determine the most suitable treatment options and inform you about eligibility for clinical trials.
- ▶ Some new DMD therapies are designed for specific genetic changes. A genetic diagnosis can tell you if one of these therapies may be appropriate for you. (Learn more about [accessing approved treatments](#).)
- ▶ A genetic diagnosis often is required to be eligible for clinical trials and other research options.
- ▶ Genetic confirmation can help determine other family members' risk for DMD.



Genetic testing can be paid for in the following ways:

- ▶ Covered by insurance
- ▶ Covered by a company that supports treatment discovery for DMD
- ▶ Self-paid

More educational resources at mda.org/education:

- ▶ [How Genetic Neuromuscular Disease Happens](#)
- ▶ [Genetics and Neuromuscular Disease](#)
- ▶ [MDA Access Workshops](#)



MDA's Resource Center may be able to help you find resources for genetic testing. Contact the Resource Center at [833-ASK-MDA1](tel:833-ASK-MDA1) or resourcecenter@mdausa.org.