

A guide to genetic testing in Duchenne muscular dystrophy

Genetic testing is a critical step in confirming a Duchenne diagnosis. Knowing your specific genetic mutation and understanding what it may mean for treatment options is key in managing the disease.

Duchenne.com



Duchenne muscular dystrophy

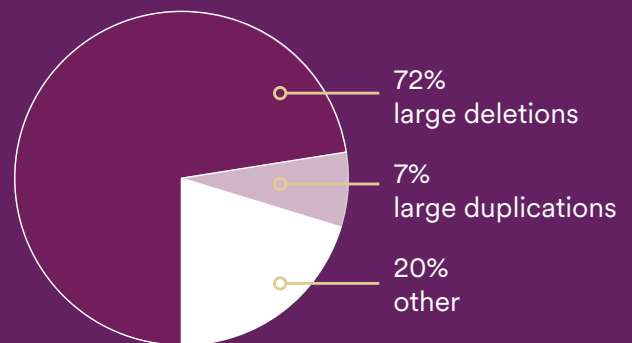


Duchenne muscular dystrophy, sometimes shortened to DMD or just Duchenne, is a rare genetic disease. Duchenne is caused by a genetic mutation, or change, in the dystrophin gene. **This mutation prevents the body from producing enough or any dystrophin, a protein that muscles need to work properly.** Without dystrophin, muscle cells become damaged and weaken over time. This change in the person's gene can either be inherited or occur spontaneously.

There are many different types of mutations of the dystrophin gene. In fact, scientists have recorded more than 7,000 unique mutations in people with the Duchenne and Becker forms of muscular dystrophy.

The most common mutation types include:

- Large deletions
- Large duplications
- Small changes



A genetic test is needed to find out what specific mutation you have. Using the genetic test results, you and your doctor can discuss care options.

Genetic testing

Genetic testing for Duchenne involves using specific laboratory methods to look at the dystrophin gene for any changes that might prevent it from working properly.

Genetic testing usually requires a blood or saliva sample.



How it works:

The goal of a genetic test is to identify change in the gene that may cause disease. Genetic testing for Duchenne should look for deletions, duplication and small changes in the *DMD* gene.



How long it takes:

Timing varies depending on which method a lab uses and whether more than one method is needed to identify the mutation. A genetic test result is usually available in approximately 21 days (3 weeks).



Who it's for:

Genetic testing can help the majority of people with Duchenne better understand their disease. Comprehensive genetic testing can detect mutations in about 95% of patients. That means that some patients (approximately 5%) may need additional testing, or may not be able to learn their mutation. Additionally, genetic testing is important for family members to understand if they too may be carriers.

Accessing genetic testing

In Duchenne, a genetic test can:



Confirm a diagnosis



Identify the genetic mutation to support consideration of care options



Provide information for identifying appropriate clinical trials for potential participation



Assist with family planning

The process for getting a genetic test generally involves a few steps:



Request genetic testing through your doctor



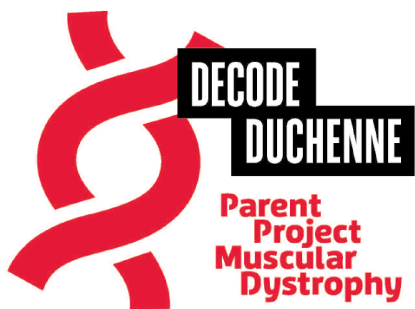
Saliva or swab kit will be sent to your home for sample collection and returned to lab for processing



Discuss your results with your doctor or genetic counselor

Genetic testing programs

There are several no-charge genetic testing programs:



Decode Duchenne:

provides no-charge genetic testing, interpretation, and counseling through PPMD and Perkin Elmer.



Detect MD:

provides no-charge genetic testing, interpretation, and counseling through Invitae.



Speak with your child's doctor about genetic testing to find out which genetic testing option is right for you and your family.

Understanding your genetic test

Every lab has a different way of reporting genetic testing results, so it can be confusing to try to understand what they mean. Your doctor should be able to discuss your test results with you. In addition, genetic counselors can work with you and your care team to:



Help you understand
the genetic
cause of Duchenne



Navigate
genetic testing,
including
understanding
the results



Provide guidance
on genetic issues
related to family
planning



Refer you
to community or
state support
services, as
appropriate



To learn more about genetic counseling or to find a genetic counselor near you, visit the National Society of Genetic Counselors (NSGC) website at aboutgeneticcounselors.com.



Knowing your specific genetic mutation provides valuable information to help you determine what disease management strategies may be appropriate, as well as whether you might be eligible for any clinical trials. Speak to your doctor or genetic counselor about your genetic testing results.

Helpful resources

Decode Duchenne

A program providing free genetic testing, interpretation and counseling to people with Duchenne or Becker muscular dystrophy who meet certain eligibility criteria and who have been unable to access genetic testing in the past due to financial barriers. Decode Duchenne is administered by DuchenneConnect, a program of Parent Project Muscular Dystrophy.

Detect MD

A program available to individuals located in the US and Canada who have a family history of muscular dystrophy or who are suspected of having muscular dystrophy.

Duchenne.com

Website that provides useful information about Duchenne muscular dystrophy, including topics like genetic testing, clinical trial participation and the drug development process. Duchenne.com is a website developed by Sarepta Therapeutics.

Genetics Home Reference

ghr.nlm.nih.gov

An online resource from the National Institutes of Health that provides information about genetic conditions. You'll find basic explanations of how genes work and how mutations cause disorders as well as information about genetic testing, gene therapy and the Human Genome Project.

National Society of Genetic Counselors

aboutgeneticcounselors.com

An online resource from the National Society of Genetic Counselors to educate patients about the role of genetic counselors. The site also includes an online directory to help you find genetic counselors in specific geographic areas.

Please talk to your doctor about other available resources.

Learn more about the
importance of
genetic testing in Duchenne.

Duchenne.com



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