Learn more about the importance of genetic testing in Duchenne at **Duchenne.com**.

If you already have your genetic test results and know the specific mutation is an exon deletion, there is an educational tool that can help you prepare for a discussion with your doctor or genetic counselor.

Visit duchenne.com/exon-deletion-tool to learn more.

Duchenne.com

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

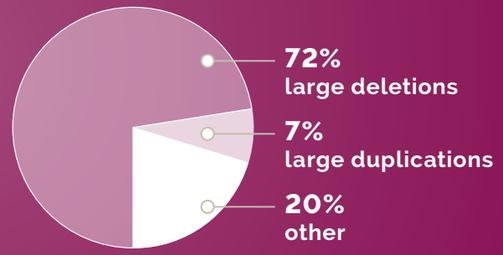
Duchenne muscular dystrophy

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 1,800 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. Sometimes, a muscle biopsy is necessary to get a genetic result, but that is not typical.

How it works:

The goal of a genetic test is to identify changes in a gene that may cause a disease. In Duchenne, laboratory scientists begin by first looking for the most common type of mutations, which are large deletions or large duplications. If a deletion or duplication is not found, another method called sequencing is done to look for small mutations.

How long it takes:

It may take from a few weeks to a few months to receive genetic test results; the timing varies depending on which method a lab uses and whether more than one method is needed to identify the mutation.

Who it's for:

Genetic testing can help the majority of people with Duchenne better understand their disease. Together, the two main methods of 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.

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

Visit a local lab to provide a blood or a saliva sample

Discuss your results with your doctor or genetic counselor

Parent Project Muscular Dystrophy (PPMD), with the support of Sarepta Therapeutics and other sponsors, has launched **Decode Duchenne**, a nationwide initiative to offer free genetic testing, interpretation, and counseling for people with Duchenne or Becker muscular dystrophy.

Decode Duchenne can provide free testing to those who:

- Have a confirmed or suspected diagnosis of Duchenne or Becker muscular dystrophy
- Have financial barriers to receiving genetic testing
- Are citizens or legal residents of the United States or Canada

The program offers:

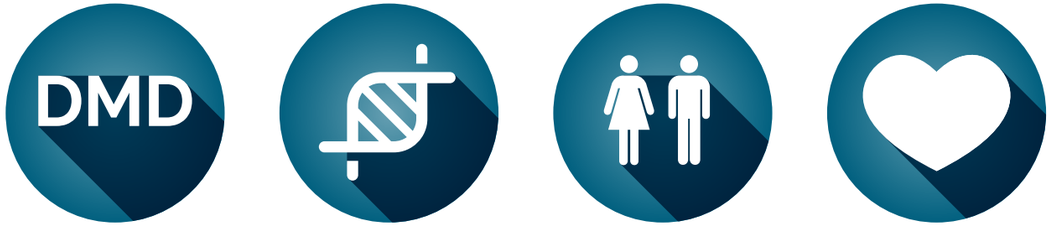
- Deletion/duplication testing with reflex to sequencing of the dystrophin gene for individuals who have never had testing
- Sequencing for those who have already had negative deletion/duplication testing and familial mutation testing
- Targeted or repeat testing for individuals tested with older technologies that did not analyze all exons in the dystrophin gene

Genetic counselors are available to walk you through the process and help determine which tests are right for you, regardless of whether you participate in the Decode Duchenne program.

For more information:
duchenneconnect.org
coordinator@duchenneconnect.org
1-888-520-8675

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

National Society of Genetic Counselors

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. **The Duchenne.com exon deletion tool, found at duchenne.com/exon-deletion-tool, may help you prepare for this discussion.**

Helpful resources

Decode Duchenne

duchenneconnect.org/decode

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.

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.