Doctor Discussion Guide

You're taking a great step forward by informing yourself and preparing, setting your child up for success in developing a personalized plan that's right for them.

It can be challenging to remember all the right questions to prepare for your child's doctor's appointment, so we've created this guide to make the process a little easier. Our guide will help address important issues about diagnosis, genetic testing, and treatment options.

IF YOU DON'T KNOW IF YOUR CHILD HAS MUSCULAR DYSTROPHY

Talk with your child's doctor about creatine kinase (CK) testing

If you suspect that your child has a muscle disease, ask your child's doctor about creatine kinase (CK) testing. Be prepared to discuss your child's development and any symptoms you've observed. If your child's doctor suspects a muscle disease, they may recommend the CK test—a simple, inexpensive laboratory test that measures the amount of creatine kinase in the blood. Elevated CK levels often indicate muscle damage and may help diagnose Duchenne muscular dystrophy.

- **Visit Duchenne.com to learn more about CK testing**
  www.duchenne.com/understanding-duchenne/diagnosing-duchenne

Have a conversation about genetic testing

Following an elevated CK, it's a great idea to schedule an appointment with a neurologist or neuromuscular specialist and talk about genetic testing.

A genetic test will pinpoint the genetic cause or specific mutation responsible for the elevated CK.

A doctor can help advise you about which treatments are appropriate for your child (also called amenability), as the right choice will vary based on your child's specific mutation.

IF YOUR CHILD DOESN'T HAVE GENETIC TEST RESULTS

Where to find genetic testing
Decode Duchenne and Detect Muscular Dystrophy both offer free genetic testing and can connect you and your doctor with genetic counselors to help interpret test results.

- **Visit Decode Duchenne to learn more**
  www.parentprojectmd.org/about-duchenne/decode-duchenne

- **Visit Detect Muscular Dystrophy to learn more**
  www.invitae.com/en/detect-muscular-dystrophy

IF YOUR CHILD ALREADY HAS GENETIC TEST RESULTS

Be sure you understand your child's genetic test results
Understanding your child's results will help you and your provider optimize the best treatment plan. Ask your child's doctor to revisit the results or consider reviewing the results with a specialized provider called a genetic counselor.

6 important questions to ask

1. Did the genetic test confirm a Duchenne diagnosis?
2. Did the test identify the genetic mutation causing my child's Duchenne?
3. What type of mutation in the DMD gene did the test show?
4. If it was a deletion, can you tell me the deletion range?
5. Is there an exon-skipping therapy appropriate for my child?
6. Should my other family members be tested?

Ask if your child's mutation is right for exon-skipping therapies
There's a good chance genetic testing may have identified the mutation causing your child's Duchenne. If you haven't already, talk with your doctor about whether your child's mutation is a good match (also called amenable) for exon-skipping therapies.

Keep the conversation going
It's important to stay up-to-date as more treatment options become available with new research. Your doctor can review your previous test results, explain their meaning, and confirm if more testing is needed.

New treatment options may have become available for some patients.

SAREPTA THERAPEUTICS