

Genetic Testing Options for Limb-girdle Muscular Dystrophy

The limb-girdle muscular dystrophies (LGMD) are a diverse group of muscle disorders with many subtypes that are categorized by the disease-causing gene and the inheritance pattern. Symptoms of LGMD include weakness and wasting (atrophy) of the muscles around the hips and shoulders. The age of onset, disease progression, and other organ involvement vary depending on the subtype.



Receiving an LGMD diagnosis

A diagnosis of LGMD is made based upon a clinical evaluation after obtaining a detailed medical history and identifying the common symptoms, including muscle weakness and atrophy. Along with the clinical evaluation, a physician may request additional testing such as an electromyography (EMG) study to look at the health of the muscles and associated nerves, specific blood tests to look at levels

of certain markers of muscle health, and/or a muscle biopsy to look at changes in muscle tissue that indicate a disease.

Genetic testing has become more routinely used to obtain a diagnosis for a patient with characteristic features of LGMD. Pursuing genetic testing is a personal decision that you should discuss with your doctor and genetic counselor to determine if it is appropriate for you.

Benefits of genetic testing

There are several reasons why genetic testing is beneficial:

- Testing can help provide an accurate diagnosis and end the diagnostic odyssey.
- There may be opportunities to participate in clinical trials focused on specific genetic subtypes of LGMD. A genetic diagnosis is a critical requirement to be able to participate in these studies.
- Having an accurate diagnosis through genetic testing can help to guide management of the disease and ensure you get the most effective treatment.
- There are several types of LGMD, and symptoms can vary depending on the type. Some types can affect the heart or the ability to breathe, and it is important to know your type so you can see the appropriate healthcare providers.
- Testing can provide information that you can use to make informed decisions about family planning. It can also help to identify other family members who could be at risk for developing symptoms of LGMD.



It may not be possible to determine every genetic cause of LGMD and some tests may come back as “inconclusive,” but as technology changes and new genetic causes are discovered, identification may become possible. It is important to

check in with your doctor every few years to see if you should be retested.



To learn more about Limb-girdle muscular dystrophy, visit mda.org/disease/limb-girdle-muscular-dystrophy or contact the MDA Resource Center at 833-ASK-MDA1.

Cost of genetic testing

Several genetic tests for LGMD are currently available from commercial laboratories and through sponsorship programs, meaning a company sponsors the test so it is free of charge to the individual. (See Sponsored Genetic Testing Programs below.) There may be eligibility requirements for sponsored testing. If you are not eligible for a sponsored program, insurance can be billed for the testing. Coverage of the cost of genetic testing depends on the insurance plan.

Genetic testing can be ordered through MDA Care Centers or neuromuscular disease physicians. Genetic counselors are also a great resource to help families understand and interpret test results. To find a genetic counselor, visit findageneticcounselor.nsgc.org.

Talk with your neurologist and a genetic counselor to determine what type of genetic test is appropriate for you.



Sponsored genetic testing programs

- **Invitae Detect Muscular Dystrophy**
- **Lantern Project: Limb-Girdle Muscular Dystrophy and Overlapping Myopathies**
- **LGMD Rare Genomes Project**
- **Jain Foundation dysferlin-specific sequencing**