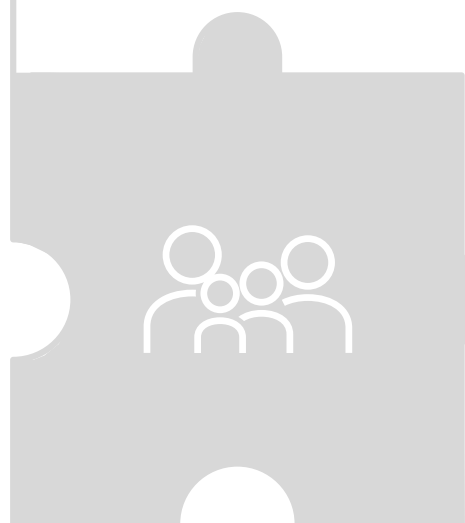




A Guide to Genetic Testing for LGMD

WHO

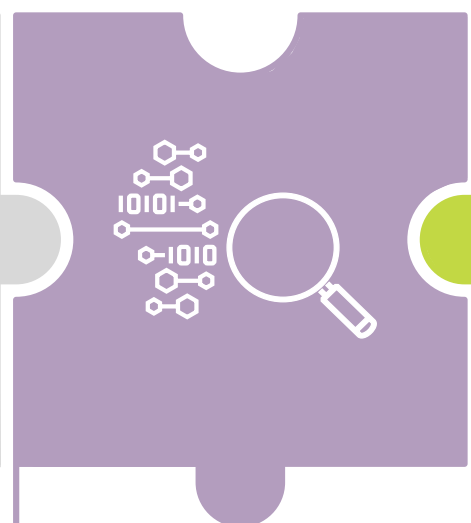
Genetic testing is **recommended for individuals with symptoms** of limb-girdle weakness that suggest LGMD, such as difficulties walking, rising to stand, raising the arms, falling and others.



A genetic test may confirm a **clinical diagnosis**—that is, one based solely on patient symptoms and medical history. Advances in testing technology means even those with prior, inconclusive genetic test results **may wish to consider getting re-tested now**.

WHAT

Most LGMD panel tests analyze the **30+ genes** associated with **LGMD subtypes** (plus sometimes genes for other muscle diseases) to look for **gene variants** (changes) that may be disease-causing.



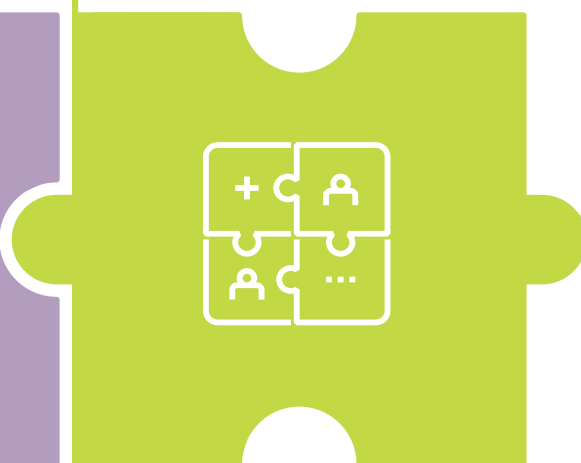
What Does “Diagnosis” Really Mean?

“LGMD” alone isn’t a diagnosis—it’s a broad disease category of 30+ separate subtypes. Genetic testing is the **only approach that may conclusively diagnose a specific subtype**, by identifying a known disease-causing gene variant.

WHY

A genetically confirmed subtype diagnosis opens up new options to:

- Work with doctor to create **personalized patient care plan** based on subtype



- Discuss with a doctor the possibility of participating in **LGMD clinical trials**, which generally require a genetic subtype diagnosis as a first step in possible patient eligibility
- Understand wider **family risk**, testing, and planning
- Connect with others in **subtype-specific LGMD communities** and advocacy organizations

LimbGirdle

Sign up for updates on **LGMD news, research, and community resources** at limbgirdle.com/stay-connected

Helpful Resources

- Find a **genetic counselor** at findageneticcounselor.nsgc.org
- Search for **LGMD clinical trials** at clinicaltrials.gov
- Consider **genetic data-sharing** at genomeconnect.org

Today, **genetic testing is accessible** to many people and considered a first-line approach to diagnosing limb-girdle muscular dystrophy (LGMD) or other similar muscle diseases.

HOW

Talk to your doctor about genetic testing to learn more. **The testing process is typically straightforward:**



- Work with a doctor or genetic counselor to determine the appropriate test and coordinate collecting a **DNA sample (typically blood or saliva)** and sending it to the lab
- Lab analyzes DNA for **gene variants** that may be disease-causing
- Lab provides **test report** within **~2-5 weeks** in U.S.

Afterwards, it's important to **review results** with a **doctor and/or genetic counselor**.

POSSIBLE TEST RESULTS

Conclusive

Close to half of those tested for suspected LGMD get a **definitive subtype diagnosis**.



Uncertain

Roughly half do not get a diagnosis because testing finds **variants of uncertain significance (VUS)**: not enough data to determine if a variant is disease-causing or not.

Negative

Minority get a negative result, suggesting that there may be more LGMD-associated genes to be discovered or that the condition involves LGMD-like muscle weakness but is not LGMD.



TAKE ACTION

Uncertain results are not the end of the diagnostic process. Patients and doctors can take follow-up actions collaboratively with the lab to collect more data, which over time may help clarify the variant.

SEEK SUPPORT

Genetic counselors are a **valuable resource** to help **interpret test results, plan next steps, and provide support throughout the testing process**.

VISIT LIMBGIRDLE.COM

Learn more about genetic testing for LGMD or other similar muscle diseases.