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.com

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.

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