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MADISON NEUROLOGY LLC

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Neurology Care 7 days a Week. Because Your Brain Matters.

Limb-Girdle Muscular Dystrophy (Adult-Onset Forms)

Patient education guide · Nerve & Muscle · Madison Neurology LLC

What is it?

Limb-girdle muscular dystrophies (LGMDs) are a group of inherited muscle diseases causing progressive weakness of the shoulder and hip muscles — difficulty lifting arms overhead, rising from chairs, and climbing stairs — usually beginning in adolescence or adulthood. Over 30 genetic subtypes exist, with widely varying severity and pace.

How does it happen?

Each subtype is caused by a mutation in a gene encoding a muscle structural or repair protein (calpain, dysferlin, sarcoglycans, FKRP, and others). Inheritance may be recessive (most common — both parents carry a silent copy) or dominant. Precise genetic diagnosis matters: some subtypes affect the heart or breathing muscles and need specific surveillance, and gene-specific trials are expanding.

Common symptoms

- Progressive hip and shoulder girdle weakness — trouble rising, stairs, overhead tasks
- Walking changes: waddling gait, frequent tripping, difficulty with inclines
- Muscle aches and cramps; high CK (muscle enzyme) often found incidentally on blood tests
- Gradual progression over years to decades
- In certain subtypes: heart muscle or rhythm involvement, breathing muscle weakness, or calf enlargement

How is it diagnosed?

Elevated CK prompts evaluation; EMG and muscle MRI characterize the pattern; genetic testing (large panels) now identifies the subtype in most patients and has largely replaced biopsy. Because some subtypes involve the heart and breathing, cardiac evaluation (ECG, echocardiogram) and breathing

tests follow diagnosis.

How is it treated?

No approved disease-modifying therapy exists yet for most subtypes, but gene therapies are advancing (an approved gene therapy exists for one subtype, with trials for others). Care is proactive: physical therapy to maintain flexibility and safe strength (avoiding overexertion), orthopedic management of contractures, mobility aids when helpful, cardiac and respiratory surveillance with early treatment of problems, and weight management. Genetic counseling guides family planning; registries connect patients to trials.

What does the future hold?

Prognosis varies enormously by subtype — from mild, near-normal lifespan with slow progression, to earlier wheelchair use and cardiopulmonary involvement. Most adults with LGMD live full lives; the cardiac and breathing surveillance catches treatable complications early. Gene-targeted therapies are the most active frontier in neuromuscular medicine.

Questions patients commonly ask

Q: Which subtype do I have, and why does it matter?

A: Genetic testing usually answers this; the subtype predicts pace, whether the heart and breathing need monitoring, and which clinical trials apply to you.

Q: Should I exercise?

A: Moderate, submaximal exercise is safe and beneficial; avoid exercising to exhaustion or heavy eccentric (lengthening) loads.

Q: What does this mean for my children?

A: It depends on inheritance type — recessive forms usually mean children are carriers at most; a genetic counselor maps your family's exact risks.

National support groups & resources

- **Muscular Dystrophy Association** — mda.org
- **National Organization for Rare Disorders** — rarediseases.org — 1-800-999-6673
- **American Academy of Neurology — Brain & Life patient resources** — brainandlife.org

(Organization contact details were current at the time of compilation; please verify online.)

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