



LIMB-GIRDLE MUSCULAR DYSTROPHY

What is Limb-Girdle Muscular Dystrophy?

Limb-Girdle Muscular Dystrophy (LGMD) is a group of hereditary disorders that cause progressive muscle weakness and wasting of the shoulders and pelvis (hips). There are at least 13 different genes that cause LGMD, each associated with a different subtype. Depending on the subtype of LGMD, the age of onset is variable (childhood, adolescence, or early adulthood) and can affect other muscles of the body. Many persons with LGMD eventually need the assistance of a wheelchair, and currently there is no cure.

How is LGMD inherited?

LGMD can be inherited by autosomal dominant (AD) or autosomal recessive (AR) modes. The AR subtypes are much more common than the AD types. Of the AR subtypes, LGMD2A (calpain-3) is the most common (30% of cases). LGMD2B (dysferlin) accounts for 20% of cases and the sarcoglycans (LGMD2C-2F) as a group comprise 25%-30% of cases. The various subtypes represent the different protein deficiencies that can cause LGMD.

What testing is available for LGMD?

Diagnosis of the LGMD subtypes requires biochemical and genetic testing. This information is critical, given that management of the disease is tailored to each individual and each specific subtype. Establishing the specific LGMD subtype is also important for determining inheritance and recurrence risks for the family.

The first step in diagnosis for muscular dystrophy is usually a muscle biopsy. Microscopic and protein analysis of the biopsy can often predict the type of muscular dystrophy by analyzing which protein(s) is absent. A muscle biopsy will allow for targeted analysis of the appropriate LGMD gene(s) and can rule out the diagnosis of the more common dystrophinopathies (Duchenne and Becker muscular dystrophies).

ChildLab is working to offer sequence analysis of the genes for the most common subtypes of LGMD (see Table 1). We discuss all abnormal results with the ordering physician and frequently discuss additional options for more challenging cases. We are also available for pre-testing questions, as each case can be unique.

Table 1

Type	Gene Symbol	Gene Name	Full Sequencing Available at ChildLab
LGMD type 1 (Dominant)			
LGMD1B	<i>LMNA</i>	Lamin A/C	YES
LGMD type 2 (Recessive)			
LGMD2A	<i>CAPN3</i>	Calpain-3	YES
LGMD2B	<i>DYSF</i>	Dysferlin	YES
LGMD2C	<i>SGCG</i>	Sarcoglycan gamma	YES
LGMD2D	<i>SGCA</i>	Sarcoglycan alpha	YES
LGMD2E	<i>SGCB</i>	Sarcoglycan beta	YES
LGMD2F	<i>SGCD</i>	Sarcoglycan delta	YES
LGMD2I	<i>FKRP</i>	Fukutin-related protein	YES

LGMD0208

Specimen requirements

- ♥ 3-10mL peripheral blood in EDTA (purple top)
- ♥ Completed requisition with muscle biopsy information
- ♥ Signed informed consent document

Expected turn around time: approximately 4-6 weeks.

For more information: For questions regarding test availability, and sample requirements, please contact [ChildLab](#) Client Services at (614)722-5477 or (800)934-6575. For technical information, please contact the Molecular Genetics Laboratory at (614)722-2866.