

GENETIC DISEASES EVALUATION CENTER

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Test Name & Code:

- **Test Name:** Limb-Girdle 2D (SGCA Gene Sequence Analysis)
- **Test Code:** 4007

Related Diseases & Genes:

- “Muscular dystrophy, limb-girdle, type 2D” (OMIM #: 608099) || Gene: SGCA (OMIM #: 600119)
- “Adhalinopathy, primary” (OMIM #:)

Synonyms:

- LGMD2D
- ADHALIN; ADL
- DYSTROGLYCAN 2; DAG2
- “DYSTROPHIN-ASSOCIATED GLYCOPROTEIN, 50-KD”
- muscular dystrophy

Test Details:

- **Methodology:** DNA Sequencing
- **Performed:** Everyday
- **Reported:** 2-3 weeks

Specimen Details:

- **Specimen Collection:** Peripheral blood
- **Container:** Tube with EDTA
- **Specimen Volume/Amount:** > 3 ml
- **Storage/Transport Condition:** Refrigerated
- **Unacceptable Specimens/Conditions:** Serum; Frozen specimens; Hemolyzed specimens; Icterus; Lipemia

Testing Area(s):

- Neurogenetics

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