

GENETIC DISEASES EVALUATION CENTER

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Test Name & Code:

- **Test Name:** *GLUD1* Gene Sequencing
- **Test Code:** 4787

Related Diseases & Genes:

- Hyperinsulinism-hyperammonemia syndrome (OMIM #: 606762) || Gene: *GLUD1* (OMIM #: 138130)

Synonyms:

- GLUD
- GDH
- GLUTAMATE DEHYDROGENASE 1

Test Details:

- **Methodology:** Next Generation DNA Sequencing
- **Performed:** Everyday
- **Reported:** 4 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):

- Multisystem Diseases

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For yesterday, today and *tomorrows...*

