

GENETIC DISEASES EVALUATION CENTER
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Test Name & Code:

- **Test Name:** Cystinosis CTNS Gene Testing
- **Test Code:** 5746

Related Diseases & Genes:

- “Cystinosis, atypical nephropathic” (OMIM #: 219800) || Gene: CTNS (OMIM #: 606272)
- “Cystinosis, late-onset juvenile or adolescent nephropathic” (OMIM #: 219800)
- “Cystinosis, nephropathic” (OMIM #: 219800)
- “Cystinosis, ocular nonnephropathic” (OMIM #: 219800)

Synonyms:

- CYSTINOSIN

Test Details:

- **Methodology:** DNA Sequencing
- **Performed:** Everyday
- **Reported:** 4 weeks

Specimen Details:

- **Specimen Collection:** Peripheral blood
- **Container:** Tube with EDTA
- **Specimen Volume/Amount:** > 5 ml
- **Storage/Transport Condition:** Refrigerated
- **Unacceptable Specimens/Conditions:** Serum; Frozen specimens; Hemolyzed specimens; Icterus; Lipemia

Testing Area(s):

- Neurogenetics
- Inborn Errors of Metabolism

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