

GENETIC DISEASES EVALUATION CENTER
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Test Name & Code:

- **Test Name:** CYP27A1 Gene Deletion/Duplication Analysis
- **Test Code:** 5648

Related Diseases & Genes:

- CEREBROTENDINOUS XANTHOMATOSIS (OMIM #: 213700) || Gene: CYP27A1 (OMIM #: 606530)

Synonyms:

- CEREBRAL CHOLESTERINOSIS
- CYP27
- STEROL 27-HYDROXYLASE

Test Details:

- **Methodology:** MLPA
- **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):

- Neurogenetics
- Inborn Errors of Metabolism

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