

GENETIC DISEASES EVALUATION CENTER
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Test Name & Code:

- **Test Name:** Sotos *NSD1* Gene Sequencing
- **Test Code:** 4057

Related Diseases & Genes:

- Sotos syndrome 1 (OMIM #: 117550) || Gene: *NSD1* (OMIM #: 606681)

Synonyms:

- *NSD1* Deletion
- 5q35 Deletion
- del 5q35
- CEREBRAL GIGANTISM

Test Details:

- **Methodology:** DNA Sequencing
- **Performed:** Everyday
- **Reported:** 8 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):

- Dysmorphology
- Neurogenetics

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For yesterday, today and *tomorrows...*

