

GENETIC DISEASES EVALUATION CENTER

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Test Name & Code:

- **Test Name:** *HOXA1* Gene Sequence Analysis
- **Test Code:** 3496

Related Diseases & Genes:

- Athabaskan brainstem dysgenesis syndrome (OMIM #: 601536) || Gene: *HOXA1* (OMIM #: 142955)
- Bosley-Salih-Alorainy syndrome (OMIM #: 601536)

Synonyms:

- HOMEBOX A1
- HOMEBOX 1F
- "Hox-1.6, MOUSE, HOMOLOG OF"
- "lab, DROSOPHILA, HOMOLOG OF"

Test Details:

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

- Dysmorphology
- Ophthalmogenetics
- Neurogenetics

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