

GENETIC DISEASES EVALUATION CENTER

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Test Name & Code:

- **Test Name:** *TWIST1* Mutation Analysis
- **Test Code:** 3230

Related Diseases & Genes:

- Craniosynostosis, type 1 (OMIM #: 123100) || Gene: *TWIST1* (OMIM #: 601622)
- Robinow-Sorauf syndrome (OMIM #: 180750)
- Saethre-Chotzen syndrome (OMIM #: 101400)
- Saethre-Chotzen syndrome with eyelid anomalies (OMIM #: 101400)

Synonyms:

- TRANSCRIPTION FACTOR TWIST;
- TWIST

Test Details:

- **Methodology:** DNA Sequencing
- **Performed:** Everyday
- **Reported:** 2-3 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
- Neurogenetics



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