

GENETIC DISEASES EVALUATION CENTER

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Test Name & Code:

- **Test Name:** *FGFR2* sequencing
- **Test Code:** 3185

Related Diseases & Genes:

- Crouzon (OMIM #: 123500) || Gene: *FGFR2* (OMIM #: 176943)
- Apert syndrome (OMIM #: 101200)
- Pfeiffer syndrome (OMIM #: 101600)
- Jackson-Weiss syndrome (OMIM #: 123150)
- Beare-Stevenson (OMIM #: 123790)

Synonyms:

-none-

Test Details:

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

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For yesterday, today and *tomorrows...*

