

GENETIC DISEASES EVALUATION CENTER
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Test Name & Code:

- **Test Name:** *FGFR1* Sequencing
- **Test Code:** 3184

Related Diseases & Genes:

- Pfeiffer syndrome (OMIM #: 101600) || Gene: *FGFR1* (OMIM #: 136350)
- Jackson-Weiss syndrome (OMIM #: 123150)
- Trigonoccephaly 1 (OMIM #: 190440)
- Osteoglophonic Dysplasia (OMIM #: 166250)
- Autosomal dominant Kallmann syndrome 2 (OMIM #: 147950)

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...*

