

GENETIC DISEASES EVALUATION CENTER
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Test Name & Code:

- **Test Name:** OSTEOGENESIS IMPERFECTA (*COL1A1* Gene)
- **Test Code:** 2781

Related Diseases & Genes:

- Osteogenesis imperfecta, type I (OMIM #: 166200) || Gene: *COL1A1* (OMIM #: 120150)

Synonyms:

- Collagen Production Defect
- Ehlers-Danlos Syndrome

Test Details:

- **Methodology:** DNA Sequencing
- **Performed:** Everyday
- **Reported:** 2 weeks

Specimen Details:

- **Specimen Collection:** Peripheral blood
- **Container:** Tube with EDTA
- **Specimen Volume/Amount:** 5-10 ml
- **Storage/Transport Condition:** Refrigerated
- **Unacceptable Specimens/Conditions:** Serum; Frozen specimens; Hemolyzed specimens; Icterus; Lipemia

Testing Area(s):

- Dysmorphology

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