



## Sequence Analysis of *COL1A1* and *COL1A2* Genes

Genetics Center offers Osteogenesis Imperfecta (OI) testing by sequencing both the *COL1A1* and *COL1A2* genes.

OI is a genetic condition caused by defects in the formation of bone, characterized by fractures with minimal or absent trauma along with a range of clinical features. OI has a prevalence of approximately 6 to 7 per 100,000, is present in all racial and ethnic groups, and is inherited in a mostly autosomal dominant fashion. Approximately 90% of OI cases are caused by defects in type I collagen, which is a component of bone that provides structural stability. In individuals with OI types I-IV, 90% have mutations in either the *COL1A1* or *COL1A2* genes. A mutation in either of these genes may result in a decrease in the amount or quality of type I collagen that is produced. When type I collagen is improperly formed, the bone loses much of its strength and fractures easily. In addition to bone fractures, patients can have short stature, hearing loss, abnormal tooth development, blue sclerae, and various bony deformities.



### ***Indications for OI testing:***

- In comprehensive work-up of non-accidental trauma.
- Multiple fractures of unknown etiology.
- Confirmation of a diagnosis of OI.
- Confirmation of a specific mutation(s) for prenatal diagnosis/family planning.

### ***Benefits of Diagnosis of and Testing for OI:***

- To provide appropriate therapies, treatment and medical management if a diagnosis of OI is known.
- Diagnosis of OI can help identify proper preventive measures and provide accurate recurrence risks for families.

Our laboratory accepts peripheral blood samples for this test. (Please see our website for further details).

As we are a comprehensive center, our medical geneticists and genetic counselors are available to provide clinical evaluations, as well as genetic counseling for patients and their families.

**For further information about testing, CPT codes, and genetic patient referrals, please visit our website at [www.geneticscenter.com](http://www.geneticscenter.com) or contact us at:**

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