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**Referred by:**
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# OSTEOGENESIS IMPERFECTA CORE PANEL COMPREHENSIVE ANALYSIS REPORT

**DATE RECEIVED:** 1/2/2021 **DATE REPORTED:** 1/14/2021 **DATE COLLECTED:** 1/1/2021

| Patient Name | DOB | Patient ID # |
|---|---|---|
| Example, Patient | 2/23/2020 | X00123 |
| **Test Request** | **Specimen Type** | **Your Code** |
| Osteogenesis Imperfecta Core Panel Comprehensive | Peripheral blood | M1234567 |

**SEQUENCING RESULTS:**

| Pathogenic Variant: | Nucleotide: | Protein: | Consistent with the clinical diagnosis of: |
|---|---|---|---|
| None | None | None | *None* |

**DELETION / DUPLICATION RESULTS:**

| Pathogenic Variant: |
|---|
| None |

**RESULTS:** DNA sequencing and copy number variation (CNV) analysis of the genes on the osteogenesis imperfecta core panel revealed no disease causing variants.
Genetic counseling is recommended for the interpretation of the results.

**METHODOLOGY:** All coding exons and exon boundaries of the genes on the osteogenesis imperfecta core panel were analyzed for variations using Illumina MiSeq next generation sequencers. New and putative disease causing single nucleotide variants and small insertions/deletions were confirmed by Sanger sequencing. Additionally, coding exons were analyzed for copy number variations (CNV). CNV detection limit is typically a single exon. The analysis does not detect rearrangements that do not result in copy number variation. The significance of the variants was determined by comparison with wild type sequences, previously reported mutations, and correlation with the protein structures. Genome build GRCh37/hg19 is used. (Note: COL1A1 exon 1 and COL1A1 exon 5 are not covered by the CNV analysis).
Although DNA sequencing and CNV analysis are highly sensitive methodologies, mutation detection may not be 100%. DNA based testing is confirmatory. A negative test result reduces but does not eliminate the chance that the genes in this test are responsible for this individual's phenotype. Additionally, a genetic cause for this individual's disorder in genomic regions not included in this test cannot be excluded.

**BACKGROUND:** Connective Tissue Gene Tests offers four panel options for osteogenesis imperfecta (OI) testing utilizing NextGen sequencing technology, an OI core panel, a dominant OI panel, a recessive OI panel and a combined dominant and recessive OI panel. In addition to genes associated with the autosomal dominant and autosomal recessive forms of OI, the panels also contain genes for disorders included in the differential diagnosis of OI. Connective Tissue Gene Tests osteogenesis imperfecta core NextGen sequencing panel consists of genes associated with osteogenesis imperfecta core.

| Gene | Accession | Disorder | MIM | Inheritance |
|---|---|---|---|---|
| COL1A1 | NM_000088 | Ehlers-Danlos syndrome, arthrochalasia type, 1 (EDSARTH1)<br>Osteogenesis imperfecta, type I (OI1)<br>Osteogenesis imperfecta, type II (OI2)<br>Osteogenesis imperfecta, type III (OI3)<br>Osteogenesis imperfecta, type IV (OI4) | 130060<br>166200<br>166210<br>259420<br>166220 | AD |

- This test was developed and its performance characteristics determined by HNL Genomics (CTGT). It has not been cleared or approved by the FDA. The FDA has determined that such clearance or approval is not necessary. This test is used for clinical purposes. It should not be regarded as investigational or for research. CTGT is certified under CLIA since 2004 to perform high complexity clinical laboratory testing.

| | | | | |
|---|---|---|---|---|
| COL1A2 | NM_000089 | Ehlers-Danlos syndrome, arthrochalasia type, 2 (EDSARTH2)<br>Osteogenesis imperfecta, type I (OI1)<br>Osteogenesis imperfecta, type II (OI2)<br>Osteogenesis imperfecta, type III (OI3)<br>Osteogenesis imperfecta, type IV (OI4) | 617821<br>166200<br>166210<br>259420<br>166220 | AD |
| IFITM5 | NM_001025295 | Osteogenesis imperfecta, type V (OI5) | 610967 | AD |

For additional information, please contact HNL Genomics (CTGT) at 484-244-2900.

Sincerely yours,

Kerry Kocher Brown, Ph.D., FACMG

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