# DISEASE:
# Osteogenesis imperfecta type 4

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| **NAME:** | Osteogenesis imperfecta type 4 |
| **DESCRIPTION:** | A moderately severe form of osteogenesis imperfecta characterized by increased bone fragility and low bone mass that clinically manifests from infancy as susceptibility to bone fractures, short stature, mild to moderate scoliosis in most, gray-blue or white sclera, and dentinogenesis imperfecta. |
| **ORPHACODE:** | 216820 |
| **SYNONYMS:** | OI type 4 |
| **XREF(S):** | Orphanet<br>MeSH<br>ICD-10<br>OMIM<br>OMIM<br>OMIM<br>OMIM<br>OMIM<br>OMIM<br>OMIM<br>OMIM<br>OMIM |

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| **ANALYTE(S):** | TMEM38B<br>COL1A1<br>COL1A2<br>CRTAP<br>PPIB<br>FKBP10<br>SP7<br>SERPINF1<br>WNT1<br>SPARC |
| **CREATED:** | 13 May 2019 - 01:02 |
| **CHANGED:** | 22 Jun 2023 - 16:14 |

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# RELATED CONTENT

## Related Genetic Tests

- Osteogenesis Imperfecta (gene panel)
- Osteogenesis imperfecta / Osteoporose (gene panel)

## Related Laboratories

- Centrum Medische Genetica - UZ Gent
- Centrum Menselijke Erfelijkheid - KUL

## Related Analytes

- collagen type I alpha 1 chain
- collagen type I alpha 2 chain
- cartilage associated protein
- FKBP prolyl isomerase 10
- peptidylprolyl isomerase B
- serpin family F member 1
- Sp7 transcription factor
- secreted protein acidic and cysteine rich
- transmembrane protein 38B
- Wnt family member 1

## Related Gene Panels

- Osteogenesis Imperfecta (25 genes) - KUL
- Osteogenesis imperfecta (18 genes) - UGent
- Osteogenesis imperfecta (3 genes) - UGent
- Osteogenesis imperfecta and Osteoporosis (43 genes) - UGent

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