DISEASE:
Osteogenesis imperfecta

NAME:	Osteogenesis imperfecta
DESCRIPTION:	A rare, genetic, primary bone dysplasias characterized by increased bone fragility, low bone mass, and susceptibility to bone fractures. The clinical severity is heterogeneous.
ORPHACODE:	666
SYNONYMS:	Brittle bone disease Glass bone disease Lobstein disease OI

XREF(S):	Orphanet MeSH MedDRA ICD-10 OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM
ANALYTE(S):	TENT5A KDELR2 MBTPS2
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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

- KDEL endoplasmic reticulum protein retention receptor 2
- membrane bound transcription factor peptidase, site 2
- terminal nucleotidyltransferase 5A

Related Gene Panels

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