DISEASE:
Ehlers-Danlos/osteogenesis imperfecta syndrome

NAME:	Ehlers-Danlos/osteogenesis imperfecta syndrome
DESCRIPTION:	A rare systemic disease characterized by the association of the features of Ehlers-Danlos syndrome with those of osteogenesis imperfecta. Predominant clinical manifestations include generalized joint hypermobility and dislocations, skin hyperextensibility and/or translucency, easy bruising, and invariable association with mild signs of osteogenesis imperfecta, including short stature, blue sclera, and osteopenia or fractures.
ORPHACODE:	230857
SYNONYMS:	EDS/OI syndrome
XREF(S):	Orphanet ICD-10 OMIM OMIM
ANALYTE(S):	COL1A1 COL1A2
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RELATED CONTENT

Related Genetic Tests

- Ehlers-Danlos syndroom, EDS (gene panel)
- 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

Related Gene Panels

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