

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
SYNOMYS:	EDS/OI syndrome
XREF(S):	Orphanet ICD-10 OMIM OMIM
ANALYTE(S):	COL1A1 COL1A2
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: <http://gentest.healthdata.be/disease/1582>

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 Erfelijkhed - 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](#)