DISEASE:
Osteogenesis imperfecta type 2

NAME:	Osteogenesis imperfecta type 2
DESCRIPTION:	A lethal type of osteogenesis imperfecta (OI) characterized by increased bone fragility, low bone mass and susceptibility to bone fractures and presenting with multiple rib and long bone fractures at birth, marked deformities, broad long bones, low density skull on X-ray, and dark sclera.
ORPHACODE:	216804
SYNONYMS:	Lethal osteogenesis imperfecta OI type 2
XREF(S):	Orphanet ICD-10 OMIM OMIM OMIM OMIM

ANALYTE(S):	COL1A1 COL1A2 P3H1 CRTAP PPIB MESD
CREATED:	13 May 2019 - 01:02
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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
- mesoderm development LRP chaperone
- prolyl 3-hydroxylase 1
- peptidylprolyl isomerase B

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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