GENETIC TEST:
Osteogenesis imperfecta / Osteoporose (gene panel)

FULL NAME:	Osteogenesis imperfecta / Osteoporose (gene panel)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Mutation confirmation, Post-natal Diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA, DNA
METHOD CATEGORY:	Sequence analysis: entire coding region Deletion/duplication analysis
METHOD TECHNIQUE:	Next Generation Sequencing (NGS)
RIZIV CODE:	565493-565504
ACCREDITATION (ISO 15189):	2021-09-11 / 2026-09-10
EQA:	• Osteogenesis Imperfecta, • Osteogenesis Imperfecta

TURNAROUND TIME (MAXIMUM):	4 months
CREATED:	11 Dec 2019 - 13:27
CHANGED:	31 Jan 2023 - 14:36
URL:	https://www.cmgg.be/nl/zorgverlener/labguide/constitutioneel-genetische-aandoen…

RELATED CONTENT

Related Diseases

- B3GALT6-related spondylodysplastic Ehlers-Danlos syndrome
- Bruck syndrome
- Ehlers-Danlos/osteogenesis imperfecta syndrome
- Familial osteochondritis dissecans
- High bone mass osteogenesis imperfecta
- Osteogenesis imperfecta
- Osteogenesis imperfecta type 1
- Osteogenesis imperfecta type 2
- Osteogenesis imperfecta type 3
- Osteogenesis imperfecta type 4
- Osteogenesis imperfecta type 5
- Short stature-advanced bone age-early-onset osteoarthritis syndrome

Related Laboratories

- Centrum Medische Genetica - UZ Gent

Related Analytes

- aggrecan
- alkaline phosphatase, biomineralization associated
- ADP ribosylation factor 5
- beta-1,3-galactosyltransferase 6

- BICD family like cargo adaptor 1
- bone morphogenetic protein 1
- coiled-coil domain containing 134
- collagen type I alpha 1 chain
- collagen type I alpha 2 chain
- COPI coat complex subunit beta 2
- cAMP responsive element binding protein 3 like 1
- cartilage associated protein
- FKBP prolyl isomerase 10
- interferon induced transmembrane protein 5
- KDEL endoplasmic reticulum protein retention receptor 2
- LIF receptor subunit alpha
- LDL receptor related protein 5
- LDL receptor related protein 6
- membrane bound transcription factor peptidase, site 2
- mesoderm development LRP chaperone
- MIA SH3 domain ER export factor 3
- NBAS subunit of NRZ tethering complex
- prolyl 3-hydroxylase 1
- prolyl 4-hydroxylase subunit beta
- phosphate regulating endopeptidase X-linked
- procollagen-lysine,2-oxoglutarate 5-dioxygenase 2
- plastin 3
- peptidylprolyl isomerase B
- SEC16 homolog B, endoplasmic reticulum export factor
- SEC24 homolog D, COPII coat complex component
- serpin family F member 1
- serpin family H member 1
- sphingomyelin synthase 2
- Sp7 transcription factor
- secreted protein acidic and cysteine rich
- syntaxin 18
- SUN domain containing ossification factor

- transmembrane anterior posterior transformation 1
- terminal nucleotidyltransferase 5A
- transmembrane protein 38B
- Wnt family member 1
- Wnt family member 3A
- xylosyltransferase 2

Related Gene Panels

- Osteogenesis imperfecta and Osteoporosis (43 genes) - UGent

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