Belgian Genetic Tests database

ANALYTE:
COL2A1

NAME:	collagen type II alpha 1 chain
SYMBOL:	COL2A1
VERSION OF ORPHANET:	2023-06-22 14:14:43
SYNONYMS:	STL1
XREF(S):	Orphanet Ensembl Genatlas HGNC OMIM Reactome SwissProt
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RELATED CONTENT

Related Genetic Tests

- Achondrogenesis / Kniest dysplasia / Hypochondrogenesis
- Cataract (gene panel)
- Congenital malformation (gene panel - 1721 genes)
- Congenital malformation gene panel
- Congenital structural heart defects (gene panel)
- Hearing loss (deafness), (gene panel)
- Intellectual disability (virtual gene panel)
- Multiple epiphyseal dysplasia
- Myopia (early onset high myopia)
- Retinal dystrophy / RETNET (gene panel)
- Skeletal dysplasia (gene panel)
- Skeletal dysplasia (gene panel)
- Skeletal dysplasia (gene panel)
- Spondylo-epiphyseal dysplasia
- Stickler syndrome (gene panel)
- Stickler syndrome (gene panel)
- Stickler syndrome (gene panel)
- cleft lip with/whitout cleft palate (virtual gene panel)

Related Diseases

- Achondrogenesis type 2
- Autosomal dominant otospondylomegaepiphyseal dysplasia
- Autosomal dominant rhegmatogenous retinal detachment

- Dysspondyloenchondromatosis
- Familial avascular necrosis of femoral head
- Hypochondrogenesis
- Kniest dysplasia
- Legg-Calvé-Perthes disease
- Mild spondyloepiphyseal dysplasia due to COL2A1 mutation with early-onset osteoarthritis
- Multiple epiphyseal dysplasia, Beighton type
- Platyspondylic dysplasia, Torrance type
- Spondyloepimetaphyseal dysplasia congenita, Strudwick type
- Spondyloepiphyseal dysplasia congenita
- Spondyloepiphyseal dysplasia with metatarsal shortening
- Spondyloepiphyseal dysplasia, Stanescu type
- Spondylometaphyseal dysplasia, 'corner fracture' type
- Spondylometaphyseal dysplasia, Schmidt type
- Spondyloperipheral dysplasia-short ulna syndrome
- Stickler syndrome type 1
- Weissenbacher- Zweymuller syndrome

Related Gene Panels

- Cataract - UGent
- Cleft lip and palate / dysmorphic facial features / craniofacial anomalies (255 genes)) - UCL
- Congenital malformation (1721 genes) - ULB
- Congenital malformation gene panel - VUB
- Congenital structural heart defects - UGent
- Hearing loss (deafness) (genepanel) - UZA
- Hearing loss (deafness) syndromic (59 genes) - UZA
- Intellectual disability (gene panel)
- Myopia gene panel - UGent
- Retinal dystrophy - UGent
- Skeletal dysplasia (394 genes) - VUB
- Skeletal dysplasia (genepanel) - UZA
- Skeletal dysplasia - UGent

- Stickler (6 genes) - KUL
- Stickler syndrome (4 genes) - UZA
- Stickler syndrome - UGent
