Stickler syndroom en gerelateerde aandoeningen genpanel

v3.0 (16 genen, incl. 6 core-NL genen)

Amsterdam UMC
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Gene	% covered > 30x	Associated phenotype description and OMIM ID
BMP4	>99	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
COL11A1	>99	Fibrochondrogenesis 1, 228520 Marshall syndrome, 154780 Stickler syndrome, type II, 604841 {Lumbar disc herniation, susceptibility to}, 603932
COL11A2	>99	Deafness, autosomal dominant 13, 601868 Deafness, autosomal recessive 53, 609706 Fibrochondrogenesis 2, 614524 Otospondylomegaepiphyseal dysplasia, 215150 Stickler syndrome, type III, 184840 Weissenbacher-Zweymuller syndrome, 277610
COL2A1	>99	Achondrogenesis type II or hypochondrogenesis, 200610 Avascular necrosis of the femoral head, 608805 Czech dysplasia, 609162 Epiphyseal dysplasia multiple with myopia and deafness, 132450 Kniest dysplasia, 156550 Legg-Calve-Perthes disease, 150600 Osteoarthritis with mild chondrodysplasia, 604864 Otospondylomegaepiphyseal dysplasia, 215150 Platyspondylic skeletal dysplasia Torrance type, 151210 SED congenita, 183900 SMED Strudwick type, 184250 Spondyloepiphyseal dysplasia Stanescu type, 616583 Spondyloperipheral dysplasia, 271700 Stickler sydrome type I nonsyndromic ocular, 609508 Stickler syndrome type I, 108300 Vitreoretinopathy with phalangeal epiphyseal dysplasia
COL9A1	>99	Stickler syndrome, type IV, 614134 ?Epiphyseal dysplasia, multiple, 6, 614135
COL9A2	>99	?Stickler syndrome, type V, 614284 Epiphyseal dysplasia, multiple, 2, 600204 {Intervertebral disc disease, susceptibility to}, 603932
COL9A3	>99	Epiphyseal dysplasia, multiple, 3, 600969 Epiphyseal dysplasia, multiple, with myopathy {Intervertebral disc disease, susceptibility to}, 603932
GZF1	>99	Joint laxity, short stature, and myopia, 617662
LOXL3	>99	no OMIM phenotype
LRP2	>99	Donnai-Barrow syndrome, 222448
P3H2	>99	Myopia, high, with cataract and vitreoretinal degeneration, 614292
PLOD3	>99	Lysyl hydroxylase 3 deficiency, 612394
SLITRK6	>99	Deafness and myopia, 221200
SLC26A2	>99	Achondrogenesis Ib, 600972 Atelosteogenesis II, 256050 De la Chapelle dysplasia, 256050 Diastrophic dysplasia, 222600 Diastrophic dysplasia broad bone-platyspondylic variant, 222600 Epiphyseal dysplasia multiple, 4226900
VCAN	>99	Wagner syndrome 1, 143200
XYLT2	>99	Spondyloocular syndrome, 605822

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014

Genes in bold are core genes

OMIM release used for OMIM disease identifiers and descriptions : June 30th, 2015

"No OMIM phenotype" signifies a gene without a current OMIM association

OMIM phenotype descriptions between {} signify risk factors