Ehlers-Danlos syndromen v3.0
(20 genen, incl. 9 core-NLgenen)

Amsterdam UMC
Universitair Medische Centra

Gene	% covered > 30x	Associated phenotype description and OMIM ID
ADAMTS2	>99	Ehlers-Danlos syndrome, type VIIC, 225410
AEBP1	>99	Ehlers-Danlos syndrome, classic-like, 2, 618000
B3GALT6	>80	Ehlers-Danlos syndrome, progeroid type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B4GALT7	>99	Ehlers-Danlos syndrome with short stature and limb anomalies, 130070
C1R	>99	Ehlers-Danlos syndrome, periodontal type, 1, 130080
C1S	>99	Ehlers-Danlos syndrome, periodontal type, 2, 617174
CHST14	>99	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
COL1A1	>99	Caffey disease, 114000 Ehlers-Danlos syndrome, classic, 130000 Ehlers-Danlos syndrome, type VIIA, 130060 Osteogenesis imperfecta, type I, 166200 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type III, 259420 Osteogenesis imperfecta, type IV, 166220 {Bone mineral density variation QTL, osteoporosis}, 166710
COL1A2	>99	Ehlers-Danlos syndrome, cardiac valvular form, 225320 Ehlers-Danlos syndrome, type VIIB, 130060 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type III, 259420 Osteogenesis imperfecta, type IV, 166220 {Osteoporosis, postmenopausal}, 166710
COL3A1	>99	Ehlers-Danlos syndrome, vascular type, 130050 Polymicrogyria with or without vascular-type EDS, 618343
COL12A1	>99	?Ullrich congenital muscular dystrophy 2, 616470 Bethlem myopathy 2, 616471
COL5A1	>99	Ehlers-Danlos syndrome, classic type, 130000
COL5A2	>99	Ehlers-Danlos syndrome, classic type, 130000
DSE	>99	?Ehlers-Danlos syndrome, musculocontractural type 2, 615539
FKBP14	>99	Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss, 614557
PLOD1	>99	Ehlers-Danlos syndrome, type VI, 225400
PRDM5	>99	Brittle cornea syndrome 2, 614170
SLC39A13	>99	Spondylocheirodysplasia, Ehlers-Danlos syndrome-like, 612350
TNXB	>99	Ehlers-Danlos syndrome due to tenascin X deficiency, 606408 Vesicoureteral reflux 8, 615963
ZNF469	>99	Brittle cornea syndrome 1, 229200

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, ? is unconfirmed data