Linkeropathieën en andere EDS-gerelateerde syndromen v3.0 (16 genen)

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Gene	% covered > 30x	Associated phenotype description and OMIM ID
B3GALT6	>80	Ehlers-Danlos syndrome, progeroid type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GAT3	>99	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600
B4GALT7	>99	Ehlers-Danlos syndrome with short stature and limb anomalies, 130070
CANT1	>99	Desbuquois dysplasia 1, 251450, 617719 Epiphyseal dysplasia, multiple, 7
CHST3	>99	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095
CSGALNACT1	>99	Skeletal dysplasia, mild, with joint laxity and advanced bone age, 618870
FLNB	>99	Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721 Boomerang dysplasia , 112310 Larsen syndrome, 150250 Spondylocarpotarsal synostosis syndrome, 272460
GORAB	>99	Geroderma osteodysplasticum, 231070
GZF1	>99	Joint laxity, short stature, and myopia, 617662
KIF22	>99	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546
PIEZO2	>99	Arthrogryposis, distal, type 3, 114300 Arthrogryposis, distal, type 5, 108145 Arthrogryposis, distal, with impaired proprioception and touch, 617146
PLOD3	>99	Lysyl hydroxylase 3 deficiency, 612394
P4HA1	>99	no OMIM phenotype
SKI	>99	Shprintzen-Goldberg syndrome, 182212
XYLT1	>99	Desbuquois dysplasia 2, 615777
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, ? is unconfirmed data