Marfan syndroom en andere marfanachtige beelden v3.0 (23genen, incl. 11 core-NL genen)

Amsterdam UMC
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Gene	% covered > 30x	Associated phenotype description and OMIM ID
ABL1	>99	Congenital heart defects and skeletal malformations syndrome, 617602
BGN	>99	Meester-Loeys syndrome, 300989 Spondyloepimetaphyseal dysplasia, X-linked, 300106
CBS		Homocystinuria due to cystathionine beta-synthase deficiency, 236200
COL3A1	>99	Ehlers-Danlos syndrome, type IV, 130050
EFEMP1	>99	Doyne honeycomb degeneration of retina, 126600
FBN1	>99	Acromicric dysplasia, 102370 Aortic aneurysm, ascending, and dissection, no OMIM numeber Ectopia lentis, familial, 129600 Geleophysic dysplasia 2, 614185 Marfan lipodystrophy syndrome, 616914 Marfan syndrome, 154700 MASS syndrome, 604308 Stiff skin syndrome, 184900 Weill-Marchesani syndrome 2, 608328
FBN2	>99	Contractural arachnodactyly, congenital , 121050 Macular degeneration, early-onset, 616118
FKBP14	>99	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
LOX	>99	Aortic aneurysm, familial thoracic 10, 617168
LTBP3	>99	no OMIM phenotype
MFAP5	>99	Aortic aneurysm, familial thoracic 9, 616166
NPR2	>99	Epiphyseal chondrodysplasia, Miura type, 615923
NPR3	>99	no OMIM phenotype
PLOD1	>99	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PMEPA1	>99	no OMIM phenotype
SKI	>99	Shprintzen-Goldberg syndrome, 182212
SLC2A10	>99	Arterial tortuosity syndrome, 208050
SMAD2	>99	no OMIM phenotype
SMAD3	>99	Loeys-Dietz syndrome 3 , 3613795
TGFB2	>99	Loeys-Dietz syndrome 4, 4614816
TGFB3	>99	Arrhythmogenic right ventricular dysplasia 1, 107970 Loeys-Dietz syndrome 5, 5615582
TGFBR1	>99	Loeys-Dietz syndrome 1, 1609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800
TGFBR2	>99	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 2610168

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