Aorta en arteriële dilatatie/dissectie v3.0
(28 genen, incl. 16 core-NL genen)

Amsterdam UMC
Universitair Medische Centra

Gene	% covered > 30x	Associated phenotype description and OMIM ID
ABL1	>99	Congenital heart defects and skeletal malformations syndrome, 617602
ACTA2	>99	Aortic aneurysm, familial thoracic 6, 611788 Moyamoya disease 5 , 5614042 Multisystemic smooth muscle dysfunction syndrome, 613834
ARIH1	>99	no OMIM phenotype
BGN	>99	Meester-Loeys syndrome, 300989 Spondyloepimetaphyseal dysplasia, X-linked, 300106
COL3A1	>99	Ehlers-Danlos syndrome, type IV, 130050
COL1A1	>99	Osteogenesis imperfecta, type I,166200 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type III, 259420 Osteogenesis imperfecta, type IV, 166220 Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1, 619115
COL5A1	>99	Ehlers-Danlos syndrome, classic type, 1, 130000
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
FOXE3	>99	{Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349
LMOD1	>99	no OMIM phenotype
LOX	>99	Aortic aneurysm, familial thoracic 10, 617168
LTBP3	>99	no OMIM phenotype
MAT2A	>99	no OMIM phenotype
MFAP5	>99	Aortic aneurysm, familial thoracic 9, 616166
MYH11	>99	Aortic aneurysm, familial thoracic 4, 132900
MYLK	>99	Aortic aneurysm, familial thoracic 7, 613780
NPR3	>99	no OMIM phenotype
PMEPA1	>99	no OMIM phenotype
PRKG1	>99	Aortic aneurysm, familial thoracic 8, 615436
SLC2A10	>99	Arterial tortuosity syndrome, 208050
SMAD2	>99	no OMIM phenotype
SMAD3	>99	Loeys-Dietz syndrome 3 , 3613795
SMAD4	>99	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350 Polyposis, juvenile intestinal, 174900
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