THORACIC AORTIC ANEURYSM AND/OR DISSECTION PANEL DG-5.0.0 (57 GENES)

Gene	*Twist X2 covered 10x*	*Twist X2 covered 20x*	*srWGS covered 10x*	*srWGS covered 15x*	*srWGS covered 20x*	*Associated Phenotype description and OMIM disease ID*
ABL1	100%	100%	100%	100%	99.7%	Leukemia, Philadelphia chromosome-positive, resistant to imatinib, 608232;Congenital heart defects and skeletal malformations syndrome, 617602
ACTA2	100%	100%	100%	100%	99.8%	Smooth muscle dysfunction syndrome, 613834;Aortic aneurysm, familial thoracic 6, 611788;Moyamoya disease 5, 614042
ARIH1	100%	100%	100%	100%	99.7%	
ASPH	100%	100%	100%	100%	99.7%	Traboulsi syndrome, 601552
BGN	99.9%	98.5%	97.4%	85.2%	66.2%	Meester-Loeys syndrome, 300989;Spondyloepimetaphyseal dysplasia, X-linked, 300106
CBS	88%	85.8%	100%	99.9%	99.2%	Thrombosis, hyperhomocysteinemic, 236200;Homocystinuria, B6-responsive and nonresponsive types, 236200

COL1A1	100%	100%	100%	100%	99.1%	Osteogenesis imperfecta, type II, 166210;Caffey disease, 114000;Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060;Osteogenesis imperfecta, type I, 166200;{Bone mineral density variation QTL, osteoporosis}, 166710;Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1, 619115;Osteogenesis imperfecta, type IV, 166220;Osteogenesis imperfecta, type III, 259420
COL1A2	100%	100%	100%	100%	99.7%	Osteogenesis imperfecta, type III, 259420;{Osteoporosis, postmenopausal}, 166710;Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821;Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2, 619120;Ehlers-Danlos syndrome, cardiac valvular type, 225320;Osteogenesis imperfecta, type IV, 166220;Osteogenesis imperfecta, type II, 166210
COL3A1	100%	100%	100%	99.9%	99.7%	Ehlers-Danlos syndrome, vascular type, 130050;Polymicrogyria with or without vascular-type EDS, 618343

COL5A1	100%	100%	100%	100%	99.3%	Ehlers-Danlos syndrome, classic type, 1, 130000;Fibromuscular dysplasia, multifocal, 619329
COL5A2	100%	100%	100%	100%	99.8%	Ehlers-Danlos syndrome, classic type, 2, 130010
DLG4	100%	100%	100%	100%	99.2%	Intellectual developmental disorder, autosomal dominant 62, 618793
EFEMP1	84.9%	84.9%	100%	100%	99.9%	Doyne honeycomb degeneration of retina, 126600;Cutis laxa, autosomal recessive, type ID, 620780;Glaucoma 1, open angle, H, 611276
EFEMP2	99.9%	97.3%	100%	99.9%	99.2%	Cutis laxa, autosomal recessive, type IB, 614437
ELN	100%	99.9%	100%	99.9%	99.4%	Cutis laxa, autosomal dominant, 123700;Supravalvar aortic stenosis, 185500
EMILIN1	100%	100%	100%	99.8%	99%	Neuronopathy, distal hereditary motor, autosomal dominant 10, 620080;Arterial tortuosity-bone fragility syndrome, 620908

FBN1	100%	100%	100%	100%	99.8%	Geleophysic dysplasia 2, 614185;Weill-Marchesani syndrome 2, dominant, 608328;Ectopia lentis, familial, 129600;MASS syndrome, 604308;Marfan lipodystrophy syndrome, 616914;Acromicric dysplasia, 102370;Marfan syndrome, 154700;Stiff skin syndrome, 184900
FBN2	99.2%	99.2%	100%	100%	99.7%	Macular degeneration, early-onset, 616118;Contractural arachnodactyly, congenital, 121050
FKBP14	100%	100%	100%	100%	100%	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FLNA	100%	100%	98.4%	87.4%	67%	Otopalatodigital syndrome, type II, 304120;Intestinal pseudoobstruction, neuronal, 300048;Cardiac valvular dysplasia, X-linked, 314400;?FG syndrome 2, 300321;Melnick-Needles syndrome, 309350;Terminal osseous dysplasia, 300244;Congenital short bowel syndrome, 300048;Otopalatodigital syndrome, type I, 311300;Heterotopia, periventricular, 1, 300049;Frontometaphyseal dysplasia 1, 305620

FOXE3	100%	100%	100%	99.5%	95.8%	Anterior segment dysgenesis 2, multiple subtypes, 610256;{Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349;Cataract 34, multiple types, 612968
FURIN	100%	99.7%	100%	100%	99.5%	
HCN4	100%	100%	100%	100%	98.8%	Sick sinus syndrome 2, 163800;{Epilepsy, idiopathic generalized, susceptibility to, 18}, 619521;Brugada syndrome 8, 613123
IPO8	97%	97%	100%	100%	99.7%	VISS syndrome, 619472
JAG1	100%	100%	100%	100%	99.7%	?Deafness, congenital heart defects, and posterior embryotoxon, 617992;Charcot-Marie-Tooth disease, axonal, type 2HH, 619574;Alagille syndrome 1, 118450;Tetralogy of Fallot, 187500
KANSL1	98.1%	98.1%	100%	100%	99.8%	Koolen-De Vries syndrome, 610443
LMOD1	100%	100%	100%	100%	98.9%	?Megacystis-microcolon-intestinal hypoperistalsis syndrome 3, 619362
LOX	100%	100%	100%	99.9%	99.1%	Aortic aneurysm, familial thoracic 10, 617168
LTBP3	100%	100%	100%	99.9%	98.8%	Dental anomalies and short stature, 601216;Geleophysic dysplasia 3, 617809
MAT2A	100%	100%	100%	100%	99.9%	

MED12	100%	100%	98.3%	86.8%	66.7%	Lujan-Fryns syndrome, 309520;Ohdo syndrome, X-linked, 300895;Hardikar syndrome, 301068;Opitz-Kaveggia syndrome, 305450
MFAP5	100%	100%	100%	100%	99.9%	Aortic aneurysm, familial thoracic 9, 616166
MYH11	100%	100%	100%	100%	99.4%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 2, 619351;Aortic aneurysm, familial thoracic 4, 132900;Visceral myopathy 2, 619350
MYLK	99.2%	99.2%	100%	99.9%	99.5%	Megacystis-microcolon-intestinal hypoperistalsis syndrome 1, 249210;Aortic aneurysm, familial thoracic 7, 613780
NOTCH1	99.1%	99%	100%	100%	99.2%	Adams-Oliver syndrome 5, 616028;Aortic valve disease 1, 109730
NPR2	100%	100%	100%	100%	99.6%	Epiphyseal chondrodysplasia, Miura type, 615923;Short stature with nonspecific skeletal abnormalities, 616255;Acromesomelic dysplasia 1, Maroteaux type, 602875
NPR3	100%	100%	100%	99.8%	98.5%	Boudin-Mortier syndrome, 619543
OLA1	100%	100%	100%	100%	99.7%	
PLOD1	96.5%	96.5%	100%	100%	99.4%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400

PMEPA1	100%	100%	100%	100%	98.5%	
PRKG1	95.9%	95.9%	100%	100%	99.6%	Aortic aneurysm, familial thoracic 8, 615436
ROBO4	100%	100%	100%	100%	99.3%	Aortic valve disease 3, 618496
SKI	100%	100%	100%	99.9%	98.9%	Shprintzen-Goldberg syndrome, 182212
SLC2A10	94.7%	94.7%	100%	100%	99.6%	Arterial tortuosity syndrome, 208050
SMAD2	86.7%	86.7%	100%	100%	99.7%	Loeys-Dietz syndrome 6, 619656;Congenital heart defects, multiple types, 8, with or without heterotaxy, 619657
SMAD3	100%	100%	100%	99.9%	99.4%	Loeys-Dietz syndrome 3, 613795
SMAD4	95.4%	95.4%	100%	100%	99.9%	Pancreatic cancer, somatic, 260350;Myhre syndrome, 139210;Polyposis, juvenile intestinal, 174900;Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050
SMAD6	100%	100%	100%	100%	98.6%	Aortic valve disease 2, 614823;{Radioulnar synostosis, nonsyndromic}, 179300;{Craniosynostosis 7, susceptibility to}, 617439
TBX20	100%	100%	100%	100%	99.8%	Atrial septal defect 4, 611363
TES	100%	100%	100%	100%	99.4%	
TGFB2	100%	100%	100%	100%	99.8%	Loeys-Dietz syndrome 4, 614816;Camurati-Engelmann disease 2, 606631

TGFB3	88.3%	88.3%	100%	100%	99.7%	Arrhythmogenic right ventricular dysplasia 1, 107970;Loeys-Dietz syndrome 5, 615582
TGFBR1	100%	100%	100%	100%	99.6%	{Multiple self-healing squamous epithelioma, susceptibility to}, 132800;Loeys-Dietz syndrome 1, 609192
TGFBR2	100%	100%	100%	100%	99.5%	Loeys-Dietz syndrome 2, 610168;Colorectal cancer, hereditary nonpolyposis, type 6, 614331;Esophageal cancer, somatic, 133239
THBS2	100%	100%	100%	100%	99.4%	?Ehlers-Danlos syndrome, classic-like, 3, 620865;{Lumbar disc herniation, susceptibility to}, 603932
THSD1	100%	100%	100%	100%	99.7%	?Aneurysm, intracranial berry, 12, 618734;Lymphatic malformation 13, 620244
THSD4	100%	100%	100%	100%	99.7%	Aortic aneurysm, familial thoracic 12, 619825

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.

TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
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srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : November 25th, 2024.
This list is accurate for panel version DG 5.0.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors