

DILATED CARDIOMYOPATHY PANEL¹ DG-4.4.0 (34 GENES)

<i>Gene</i>	<i>Twist X2 covered 10x</i>	<i>Twist X2 covered 20x</i>	<i>srWGS covered 10x</i>	<i>srWGS covered 15x</i>	<i>srWGS covered 20x</i>	<i>Associated Phenotype description and OMIM disease ID</i>
ACTC1	100%	100%	100%	100%	99.5%	Left ventricular noncompaction 4, 613424;Cardiomyopathy, hypertrophic, 11, 612098;Atrial septal defect 5, 612794;Cardiomyopathy, dilated, 1R, 613424
ACTN2	100%	100%	99.9%	99.3%	98.1%	Myopathy, distal, 6, adult onset, 618655;Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158;Congenital myopathy 8, 618654;Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158

BAG3	100%	100%	100%	100%	99%	?Neuronopathy, distal hereditary motor, autosomal dominant 15, 621094;Cardiomyopathy, dilated, 1HH, 613881;Myopathy, myofibrillar, 6, 612954;Charcot-Marie-Tooth disease, axonal, type 2JJ, 621095
BAG5	100%	100%	100%	99.8%	98%	Cardiomyopathy, dilated, 2F, 619747
DES	100%	100%	100%	99.6%	97.6%	Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400;Cardiomyopathy, dilated, 1I, 604765;Myopathy, myofibrillar, 1, 601419
DMD	99.6%	99.6%	99.4%	93.2%	75.9%	Becker muscular dystrophy, 300376;Cardiomyopathy, dilated, 3B, 302045;Duchenne muscular dystrophy, 310200

DSP	100%	100%	100%	100%	99.4%	Arrhythmogenic right ventricular dysplasia 8, 607450;Epidermolysis bullosa, lethal acantholytic, 609638;Keratosis palmoplantaris striata II, 612908;Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821;Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676
FLII	100%	100%	100%	99.9%	98.9%	Cardiomyopathy, dilated, 2J, 620635
FLNC	100%	100%	100%	100%	99.4%	Cardiomyopathy, familial hypertrophic, 26, 617047;Arrhythmogenic right ventricular dysplasia, familial, 617047;Cardiomyopathy, familial restrictive 5, 617047;Myopathy, distal, 4, 614065;Myopathy, myofibrillar, 5, 609524

HCN4	100%	100%	100%	99.8%	98.3%	Sick sinus syndrome 2, 163800;{Epilepsy, idiopathic generalized, susceptibility to, 18}, 619521;Brugada syndrome 8, 613123
JPH2	100%	100%	100%	100%	98.1%	Cardiomyopathy, dilated, 2E, 619492;Cardiomyopathy, hypertrophic, 17, 613873
LDB3	100%	100%	100%	100%	99.1%	Left ventricular noncompaction 3, 601493;Cardiomyopathy, dilated, 2L, 621237;Cardiomyopathy, hypertrophic, 24, 601493;Myopathy, myofibrillar, 4, 609452;Cardiomyopathy, dilated, 1C, with or without LVNC, 601493

LMNA	100%	100%	100%	100%	99.3%	Mandibuloacral dysplasia, 248370;Heart-hand syndrome, Slovenian type, 610140;Cardiomyopathy, dilated, 1A, 115200;Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516;Restrictive dermopathy 2, 619793;Charcot-Marie-Tooth disease, type 2B1, 605588;Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350;Hutchinson-Gilford progeria, 176670;Lipodystrophy, familial partial, type 2, 151660;Muscular dystrophy, congenital, 613205;Malouf syndrome, 212112
LMOD2	100%	100%	100%	99.9%	99.4%	Cardiomyopathy, dilated, 2G, 619897

MYH7	100%	100%	100%	100%	99.4%	Laing distal myopathy, 160500;Cardiomyopathy, hypertrophic, 1, 192600;Left ventricular noncompaction 5, 613426;Cardiomyopathy, dilated, 1S, 613426;Congenital myopathy 7B, myosin storage, autosomal recessive, 255160;Congenital myopathy 7A, myosin storage, autosomal dominant, 608358
MYLK3	100%	100%	100%	99.9%	99.2%	
MYZAP	100%	100%	100%	100%	99.6%	Cardiomyopathy, dilated, 2K, 620894
NEXN	100%	100%	100%	100%	99.6%	Cardiomyopathy, dilated, 2M, autosomal recessive, 621261;Cardiomyopathy, dilated, 1CC, 613122;Cardiomyopathy, hypertrophic, 20, 613876
NRAP	100%	100%	100%	100%	99.6%	
PLEKHM2	100%	100%	100%	99.9%	99.1%	
PLN	100%	100%	100%	100%	99.2%	Cardiomyopathy, dilated, 1P, 609909;Cardiomyopathy, hypertrophic, 18, 613874

PPA2	100%	100%	100%	100%	99.8%	?Sudden cardiac failure, alcohol-induced, 617223;Sudden cardiac failure, infantile, 617222
PRDM16	100%	100%	100%	99.9%	98.7%	Left ventricular noncompaction 8, 615373;Cardiomyopathy, dilated, 1LL, 615373
RBM20	100%	100%	100%	100%	99.6%	Cardiomyopathy, dilated, 1DD, 613172
RPL3L	100%	100%	100%	99.8%	98.5%	Cardiomyopathy, dilated, 2D, 619371
SCN5A	100%	100%	100%	99.9%	99.2%	Ventricular fibrillation, familial, 1, 603829;Heart block, progressive, type IA, 113900;Cardiomyopathy, dilated, 1E, 601154;Heart block, nonprogressive, 113900;Long QT syndrome 3, 603830;Sick sinus syndrome 1, 608567;Brugada syndrome 1, 601144;Atrial fibrillation, familial, 10, 614022;{Sudden infant death syndrome, susceptibility to}, 272120

TBX20	100%	100%	100%	100%	99.1%	Atrial septal defect 4, 611363
TNNC1	100%	100%	100%	100%	99.5%	Cardiomyopathy, dilated, 1Z, 611879;Cardiomyopathy, hypertrophic, 13, 613243
TNNI3	100%	100%	100%	99.7%	97.4%	?Cardiomyopathy, dilated, 2A, 611880;Cardiomyopathy, hypertrophic, 7, 613690;Cardiomyopathy, familial restrictive, 1, 115210;Cardiomyopathy, dilated, 1FF, 613286
TNNI3K	100%	100%	100%	100%	99.8%	Cardiac conduction disease with or without dilated cardiomyopathy, 616117
TNNT2	100%	100%	100%	99.9%	99.5%	Cardiomyopathy, dilated, 1D, 601494;Cardiomyopathy, hypertrophic, 2, 115195;Cardiomyopathy, familial restrictive, 3, 612422;Left ventricular noncompaction 6, 601494

TPM1	100%	100%	100%	99.9%	99.1%	Left ventricular noncompaction 9, 611878;Cardiomyopathy, hypertrophic, 3, 115196;Cardiomyopathy, dilated, 1Y, 611878
TTN	100%	100%	100%	100%	99.7%	Muscular dystrophy, limb-girdle, autosomal recessive 10, 608807;Congenital myopathy 5 with cardiomyopathy, 611705;Tibial muscular dystrophy, tardive, 600334;Cardiomyopathy, dilated, 1G, 604145;?Cardiomyopathy, familial hypertrophic, 9, 613765;Myopathy myofibrillar, 9, with early respiratory failure, 603689
VCL	100%	100%	100%	100%	99.5%	Cardiomyopathy, dilated, 1W, 611407;Cardiomyopathy, hypertrophic, 15, 613255

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.

srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.

*srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
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 4.4.0

Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors