HYPERTROPHIC CARDIOMYOPATHY PANEL[1] DG-5.2.0 (29 GENES)

Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
OMIM disease identifiers and descriptions : October 25th, 2026.
This list is accurate for panel version DG 5.2.0
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
Ad 1. Blank field signifies a gene without a current OMIM association
Ad 2. OMIM phenotype descriptions between {} signify risk factors
*Ad * This value indicates the percentage of a gene's coding sequence covered at or above the specified Genotype Quality (GQ) threshold, based on alignment against the GRCh38 reference genome. The GQ score is a measure of the confidence of the genotype call and is calculated based on both the quantity and quality of the DNA reads.*
- GQ 20: 99% confidence in the genotype call
- GQ 30: 99.9% confidence in the genotype call

Gene	WES GQ>20 | GQ>30*	srWGS GQ>20 | GQ>30*	lrWGS GQ>20 | GQ>30*	Associated Phenotype description and OMIM disease ID
ACTC1	91% | 91%	97% | 97%	100% | 100%	Left ventricular noncompaction 4, 613424;Cardiomyopathy, hypertrophic, 11, 612098;Atrial septal defect 5, 612794;Cardiomyopathy, dilated, 1R, 613424
ACTN2	98% | 97%	96% | 96%	99% | 99%	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
ALPK3	100% | 100%	96% | 96%	99% | 99%	Cardiomyopathy, familial hypertrophic 27, 618052
CACNA1C	100% | 100%	96% | 96%	99% | 99%	Timothy syndrome, 601005;Long QT syndrome 8, 618447;Neurodevelopmental disorder with hypotonia, language delay, and skeletal defects with or without seizures, 620029;Brugada syndrome 3, 611875
CSRP3	100% | 100%	97% | 97%	100% | 100%	?Cardiomyopathy, dilated, 1M, 607482;Cardiomyopathy, hypertrophic, 12, 612124
DES	92% | 92%	95% | 95%	98% | 98%	Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400;Cardiomyopathy, dilated, 1I, 604765;Myopathy, myofibrillar, 1, 601419
FHL1	98% | 98%	99% | 99%	100% | 100%	Myopathy, X-linked, with postural muscle atrophy, 300696;Emery-Dreifuss muscular dystrophy 6, X-linked, 300696;?Uruguay faciocardiomusculoskeletal syndrome, 300280;Scapuloperoneal myopathy, X-linked dominant, 300695;Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718;Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717
FHOD3	98% | 98%	97% | 97%	99% | 99%	Cardiomyopathy, familial hypertrophic, 28, 619402
FLNC	100% | 100%	97% | 97%	98% | 98%	Cardiomyopathy, familial dilated, 1PP, 617047;Cardiomyopathy, familial hypertrophic, 26, 617047;Arrhythmogenic right ventricular dysplasia, familial, 617047;Cardiomyopathy, familial restrictive 5, 617047;Myopathy, distal, 4, 614065;Myopathy, myofibrillar, 5, 609524
GLA	91% | 91%	99% | 99%	100% | 100%	Fabry disease, cardiac variant, 301500;Fabry disease, 301500
JPH2	93% | 93%	93% | 93%	99% | 99%	Cardiomyopathy, dilated, 2E, 619492;Cardiomyopathy, hypertrophic, 17, 613873

KLHL24	92% | 92%	98% | 98%	99% | 99%	Cardiomyopathy, familial hypertrophic, 29, with polyglucosan bodies, 620236;Epidermolysis bullosa simplex 6, generalized intermediate, with or without cardiomyopathy, 617294
LAMP2	71% | 71%	98% | 98%	100% | 100%	Danon disease, 300257
MT-TI				
MYBPC3	100% | 100%	96% | 96%	99% | 99%	Cardiomyopathy, hypertrophic, 4, 115197;Cardiomyopathy, dilated, 1MM, 615396;Left ventricular noncompaction 10, 615396
MYH7	99% | 99%	97% | 97%	99% | 99%	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
MYL2	100% | 100%	97% | 97%	99% | 99%	Cardiomyopathy, hypertrophic, 10, 608758;Myopathy, myofibrillar, 12, infantile-onset, with cardiomyopathy, 619424
MYL3	79% | 79%	97% | 97%	99% | 99%	Cardiomyopathy, hypertrophic, 8, 608751
PLN	100% | 100%	98% | 98%	100% | 100%	Cardiomyopathy, dilated, 1P, 609909;Cardiomyopathy, hypertrophic, 18, 613874
PRKAG2	100% | 100%	96% | 96%	99% | 99%	Glycogen storage disease of heart, lethal congenital, 261740;Wolff-Parkinson-White syndrome, 194200;Cardiomyopathy, hypertrophic 6, 600858
PTPN11	89% | 89%	98% | 98%	99% | 99%	Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondromatosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785
RAF1	93% | 93%	97% | 97%	99% | 99%	Cardiomyopathy, dilated, 1NN, 615916;Noonan syndrome 5, 611553;LEOPARD syndrome 2, 611554
RIT1	100% | 100%	97% | 97%	99% | 99%	Noonan syndrome 8, 615355
TNNC1	100% | 100%	97% | 97%	99% | 99%	Cardiomyopathy, dilated, 1Z, 611879;Cardiomyopathy, hypertrophic, 13, 613243
TNNI3	100% | 100%	95% | 95%	97% | 97%	?Cardiomyopathy, dilated, 2A, 611880;Cardiomyopathy, hypertrophic, 7, 613690;Cardiomyopathy, familial restrictive, 1, 115210;Cardiomyopathy, dilated, 1FF, 613286
TNNT2	100% | 100%	96% | 96%	99% | 99%	Cardiomyopathy, dilated, 1D, 601494;Cardiomyopathy, hypertrophic, 2, 115195;Cardiomyopathy, familial restrictive, 3, 612422;Left ventricular noncompaction 6, 601494
TPM1	91% | 91%	97% | 97%	99% | 99%	Left ventricular noncompaction 9, 611878;Cardiomyopathy, hypertrophic, 3, 115196;Cardiomyopathy, dilated, 1Y, 611878
TRIM63	100% | 100%	98% | 98%	99% | 99%	Cardiomyopathy, familial hypertrophic, 31, 621270
TTR	100% | 100%	98% | 98%	100% | 100%	Amyloidosis, hereditary systemic 1, 105210;Carpal tunnel syndrome, familial, 115430;[Dystransthyretinemic hyperthyroxinemia], 145680