|  |
| --- |
| **CARDIOMYOPATHY EXOME PANEL** |
| **Gene**  | **Inheritance** | **OMIM** | **Cardiomyopathy Associated Phenotypes** | **Other Phenotypes** |
| **HCM** | **DCM** | **ARVC** | **LVNC** | **RCM** | **CPVT** |
| ***ABCC9*** | AD | [601439](http://www.omim.org/entry/601439) |   | X |   |   |   |   | Atrial fibrillation, familial, 12 (AD); Hypertrichotic osteochondrodysplasia (AD); Cantu syndrome (AD) |
| ***ACTC1*** |  AD | [102540](http://www.omim.org/entry/102540) | X | X |   | X | X |   | Atrial septal defect 5 |
| ***ACTN2*** | AD | [102573](http://www.omim.org/entry/102573) | X | X |   | X |   |   |   |
| ***ANKRD1*** | UNKN | [609599](http://www.omim.org/entry/609599) | X | X |   |   |   |   |   |
| ***BAG3*** | AD | [603883](http://www.omim.org/entry/603883) | X | X |   |   | X |   | Myopathy, myofibrillar, 6 (AD) |
| ***CASQ2*** | AD/AR | [114251](http://www.omim.org/entry/114251) |   |   |   | X |   | X |   |
| ***CAV3*** |  AD | [601253](http://www.omim.org/entry/601253) | X | X |   |   |   |   |  Long QT syndrome 9 (AD) Myopathy, distal, Tateyama type (AD) |
| ***CHRM2*** | AD | [118493](http://www.omim.org/entry/118493) |   | X |   |   |   |   |   |
| ***CRYAB*** | UNKN | [123590](http://www.omim.org/entry/123590) |   | X |   |   |   |   | Myopathy, myofibrillar (AD/AR) |
| ***CSRP3*** | AD | [600824](http://www.omim.org/entry/600824) | X | X |   |   |   |   | Myopathy with HCM (AD) |
| ***DES*** | AD | [125660](http://www.omim.org/entry/125660) |   | X | X |   |   | X | Muscular dystrophy, limb-girdle, type 2R (AD); Myopathy, myofibrillar, 1C (AR) |
| ***DMD*** | XL | [300377](http://www.omim.org/entry/300377) |   | X |   |   |   |   | Becker muscular dystrophy; Duchenne muscular dystrophy; Female carriers may develop isolated DCM |
| ***DOLK*** | AR | [610746](http://www.omim.org/entry/610746) |   | X |   |   |   |   |   |
| ***DSC2*** | AD  | [125645](http://www.omim.org/entry/125645) |   | X | X |   |   |   |   |
| ***DSG2*** | AD  | [125671](http://www.omim.org/entry/125671) |   | X | X |   |   |   |   |
| ***DSP*** | AD  | [125647](http://www.omim.org/entry/125647) |   | X | X |   |   |   | Epidermolysis bullosa, lethal acantholytic (AR); Carvajal syndrome (AR) |
| ***DTNA*** | AD | [601239](http://www.omim.org/entry/601239) |   |   |   | X |   |   |   |
| ***EMD*** | XL | [300384](http://www.omim.org/entry/300384) |   | X |   |   |   |   | Emery-Dreifuss muscular dystrophy 1 |
| ***FHL2*** | UNKN | [602633](http://www.omim.org/entry/602633) |   | X |   |   |   |   |   |
| ***FKTN*** | AR | [607440](http://www.omim.org/entry/607440) |   | X |   |   |   |   |  Muscular dystrophy, type A4 (AR) |
| ***GAA*** | AR | [606800](http://www.omim.org/entry/606800) | X |   |   |   |   |   | Glycogen storage disease II (AR) |
| ***GATAD1*** | AR | [614518](http://www.omim.org/entry/614518) |   | X |   |   |   |   |   |
| ***GLA*** | XL | [300644](http://www.omim.org/entry/300644) | X |   |   |   |   |   | Fabry disease |
| ***ILK*** | UNKN | [602366](http://www.omim.org/entry/602366) |   | X |   |   |   |   |   |
| ***JPH2*** | AD | [605267](http://www.omim.org/entry/605267) | X |   |   |   |   |   |   |
| ***JUP*** | AD | [173325](http://www.omim.org/entry/173325) |   | X | X |   |   |   | Naxos disease (AR) |
| ***LAMA4*** | AD | [600133](http://www.omim.org/entry/600133) |   | X |   |   |   |   |   |
| ***LAMP2*** | XL | [309060](http://www.omim.org/entry/309060) | X | X |   |   |   |   | Danon disease  |
| ***LDB3*** | AD  | [605906](http://www.omim.org/entry/605906) | X | X |   | X |   |   | Myopathy, myofibrillar, 4 |
| ***LMNA*** | AD  | [150330](http://www.omim.org/entry/150330) |   | X |   | X |   |   | See OMIM for other associated phenotypes  |
| ***MURC*** | AD |   |   | X |   |   |   |   |   |
| ***MYBPC3*** | AD | [600958](http://www.omim.org/entry/600958) | X | X |   | X | X |   |   |
| ***MYH6*** | AD | [160710](http://www.omim.org/entry/160710) | X | X |   |   |   |   | Atrial septal defect 3 (AD)  |
| ***MYH7*** |  AD  | [160760](http://www.omim.org/entry/160760) | X | X |   | X | X |   | Liang distal myopathy, (AD); Myopathy, myosin storage, (AD); Myopathy, myosin storage (AR) |
| ***MYL2*** | AD | [160781](http://www.omim.org/entry/160781) | X |   |   |   |   |   |   |
| ***MYL3*** | AD | [160790](http://www.omim.org/entry/160790) | X |   |   |   | X |   |   |
| ***MYLK2*** | AD | [606566](http://www.omim.org/entry/606566) | X |   |   |   |   |   |   |
| ***MYOZ2*** | AD | [605602](http://www.omim.org/entry/605602) | X |   |   |   |   |   |   |
| ***MYPN*** | AD | [608517](http://www.omim.org/entry/608517) | X | X |   |   | X |   |   |
| ***NEBL*** | UNKN | [605491](http://www.omim.org/entry/605491) |   | X |   |   |   |   |   |
| ***NEXN*** | UNKN | [613121](http://www.omim.org/entry/613121) | X | X |   | X |   |   |   |
| ***PDLIM3*** | UNKN | [605889](http://www.omim.org/entry/605889) | X | X | X |   |   |   |   |
| ***PKP2*** | AD | [602861](http://www.omim.org/entry/602861) |   | X | X |   |   |   |   |
| ***PLN*** | AD | [172405](http://www.omim.org/entry/172405) | X | X | X |   |   |   |   |
| ***PRDM16*** | AD | [605557](http://www.omim.org/entry/605557) |   | X |   | X |   |   |   |
| ***PRKAG2*** | AD | [602743](http://www.omim.org/entry/602743) | X | X |   |   |   |   | Glycogen storage disease of heart, lethal congenital; Wolff-Parkinson-White syndrome |
| ***PSEN1*** | AD | [104311](http://www.omim.org/entry/104311) |   | X |   |   |   |   |   |
| ***PSEN2*** | AD | [600759](http://www.omim.org/entry/600759) |   | X |   |   |   |   |   |
| ***RBM20*** | AD | [613171](http://www.omim.org/entry/613171) |   | X |   |   |   |   |   |
| ***RYR2*** | AD | [180902](http://www.omim.org/entry/180902) | X |   | X |   |   | X |   |
| ***SCN5A*** | AD | [600163](http://www.omim.org/entry/600163) |   | X | X |   |   |   | Brugada syndrome 1 (AD); Long QT syndrome-3 (AD) |
| ***SGCD*** | AD | [601411](http://www.omim.org/entry/601411) |   | X |   |   |   |   | Muscular dystrophy, limb-girdle, type 2F (AR) |
| ***TAZ*** | XL | [300394](http://www.omim.org/entry/300394) |   | X |   | X |   |   | Barth syndrome  |
| ***TCAP*** | AD  | [604488](http://www.omim.org/entry/604488) | X |   |   |   |   |   | Muscular dystrophy, limb-girdle, type 2G (AR) |
| ***TMEM43*** | AD | [612048](http://www.omim.org/entry/612048) |   |   | X |   |   |   | Emery-Dreifuss muscular dystrophy 7 |
| ***TMPO*** | AD | [188380](http://www.omim.org/entry/188380) |   | X |   |   |   |   |   |
| ***TNNC1*** | AD | [191040](http://www.omim.org/entry/191040) | X | X |   |   |   |   |   |
| ***TNNI3*** | AD  | [191044](http://www.omim.org/entry/191044) | X | X |   |   | X |   |   |
| ***TNNT2*** | AD | [191045](http://www.omim.org/entry/191045) | X | X |   | X | X |   |   |
| ***TRDN*** | AR | [603283](http://www.omim.org/entry/603283) |   |   |   |   |   | X |   |
| ***TTN*** | AD | [188840](http://www.omim.org/entry/188840) | X | X | X |   |   |   | Muscular dystrophy, limb-girdle (AR); Tibial muscular dystrophy (AD/AR), HMERF (AD), Centronuclear myopathy (AR) |
| ***TTR*** | AD | [176300](http://www.omim.org/entry/176300) | X | X |   |   |   |   | Amyloidosis |
| ***VCL*** | AD | [193065](http://www.omim.org/entry/193065) | X | X |   | X |   |   |   |
| ***BRAF*** | AD | [164757](http://www.omim.org/entry/164757) | X |   |   |   |   |   | Cardiofaciocutaneous syndrome; Noonan syndrome; LEOPARD syndrome |
| ***CBL*** | AD | [165360](http://www.omim.org/entry/165360) | X |   |   |   |   |   | Noonan syndrome |
| ***HRAS*** | AD | [190020](http://www.omim.org/entry/190020) | X |   |   |   |   |   | Costello syndrome |
|  |  |  |  |  |  |  |  |  |  |
| ***KRAS*** | AD | [190070](http://www.omim.org/entry/190070) | X |   |   |   |   |   | Cardiofaciocutaneous syndrome; Noonan syndrome |
| ***NRAS*** | AD  | [164790](http://www.omim.org/entry/164790) | X |   |   |   |   |   | Noonan syndrome |
| ***PTPN11*** | AD | [176876](http://www.omim.org/entry/176876) | X |   |   |   |   |   | Noonan syndrome; LEOPARD syndrome |
| ***RAF1*** | AD | [164760](http://www.omim.org/entry/164760) | X | X |   |   |   |   | Noonan syndrome; LEOPARD syndrome |
| ***RIT1*** | AD | [609591](http://www.omim.org/entry/609591) | X |   |   |   |   |   | Noonan syndrome |
| ***SHOC2*** | AD | [602775](http://www.omim.org/entry/602775) | X |   |   |   |   |   | Noonan syndrome |
| ***SOS1*** | AD | [182530](http://www.omim.org/entry/182530) | X |   |   |   |   |   | Noonan syndrome  |
| ***MAP2K1*** | AD | 176872 | X |   |   |   |   |   | Cardiofaciocutaneous syndrome |
| ***MAP2K2*** | AD | 601263 | X |   |   |   |   |   | Cardiofaciocutaneous syndrome |

HCM = Hypertrophic cardiomyopathy; DCM = Dilated cardiomyopathy; ARVC =Arrythmogenic right ventricular dysplasia; LVNC = Left ventricular non-compaction; RCM = Restrictive cardiomyopathy; CPVT = Catecholaminergic polymorphic ventricular tachycardia