Supplemental Table 1: Missense mutations in MYH7 (β-cardiac myosin heavy chain) and their associated disease. HCM: hypertrophic cardiomyopathy. DCM: dilated cardiomyopathy. LVNC: Left Ventricular non-compaction.

Mutations	Myosin region	Disease	Reference
Ser4Leu		HCM	[1]
Gly10Ala		HCM	[2]
Gly10Arg		HCM	[3]
Ala13Thr		HCM	[4]
Arg17Cys		HCM	[5]
Arg17His		HCM	[6]
Ala23Trp		LVNC	[7]
Ala26Gly		HCM	[8]
Ala26Val		HCM	[9]
Gln27Arg		HCM	[3]
Val39Met	SH3-like fold	HCM	[10]
Asp41Asn	SH3-like fold	HCM	[11]
Gln44Term	SH3-like fold	LVNC	[12]
Glu45Asp	SH3-like fold	HCM	[3]
Phe46Cys	SH3-like fold	HCM	[3]
Phe46Ile	SH3-like fold	HCM	[13]
Val52Met	SH3-like fold	HCM	[14]
Arg54Gln	SH3-like fold	HCM	[3]
Arg54Term	SH3-like fold	HCM	[15]
Val59Ile	SH3-like fold	HCM	[8]
Glu62Lys	SH3-like fold	DCM	[3]
Thr70Ser	SH3-like fold	HCM	[16]
Pro81Ser		DCM	[17]
Ile87Thr		HCM	[14]
Asp89Gly		LVNC	[18]
Phe95Ile		HCM	[19]
Glu98Val		HCM	[3]
Ala100Thr		HCM	[20]
Ser111Phe		LVNC	[21]
Ile114Thr		HCM	[22]
Tyr115His		HCM	[23]
Tyr115Term		HCM	[6]
Thr116Ser		HCM	[13]
Tyr117Phe		HCM	[24]
Ser118Leu		HCM	[25]
Thr124Ile		HCM	[26]
Pro127Thr		LVNC	[27]
Thr135Ile		HCM	[19]
Glu137Lys		HCM	[22]
Val139Leu		DCM	[11]
Tyr142His		DCM	[17]
Arg143Gly		HCM	[28]
Arg143Trp		HCM	[29]
Arg143Gln		HCM	[30]
Gly144Asp		HCM	[24]
Gly144Val		DCM	[31]

Lys146Asn		HCM	[32]
Arg147Ser		HCM	[33]
Ser148Ile		HCM	[28]
Pro151Leu		Myopathy	[34]
Tyr162Cys		HCM	[26]
Tyr162His		HCM	[35]
Gln163Pro		LVNC	[36]
Met165Ile		LVNC	[18]
Thr167Ile		HCM	[3]
Asp168Asn		HCM	[24]
Arg169Gly		HCM	[37]
Arg169Lys		HCM	[5]
Arg169Ser		HCM	[5]
Glu170Lys		HCM	[38]
Thr177Ile		HCM	[39]
Thr177Ser		HCM	[40]
Gly178Arg	P-loop	HCM and SM	[17]
Gly181Arg	P-loop	DCM	[3]
Gly183Arg	P-loop	CM	[18]
Lys184Gln	P-loop	LVNC	[41]
Val186Leu		HCM	[32]
Asn187Lys		HCM	[26]
Asn187Ser		HCM	[24]
Thr188Asn		HCM	[10]
Arg190Thr		HCM	[42]
Gln193Arg		HCM	[3]
Tyr194Ser		HCM	[43]
Ala196Thr		HCM	[44]
Val197Ile		DCM	[3]
Ala199Thr	Loop-1	HCM	[14]
Ala199Val	Loop-1	CM	[37]
Ala200Thr	Loop-1	HCM	[45]
Ile201Thr	Loop-1	DCM	[46]
Arg204Cys	Loop-1	HCM	[24]
Arg204His	Loop-1	HCM	[10]
Arg204Leu	Loop-1	HCM	[3]
Ser205Cys	Loop-1	HCM	[47]
Lys207Gln	Loop-1	HCM	[28]
Gln209Glu	Loop-1	HCM	[47]
Gln209Lys	Loop-1	DCM	[48]
Ser210Arg	Loop-1	HCM	[25]
Pro211Leu	Loop-1	HCM	[28]
Gly214Asp	Loop-1	HCM	[49]
Leu216Val		HCM	[49]
Asp218Tyr		HCM	[19]
Gln219Leu		HCM	[50]
Gln219Glu		HCM	[50]
Gln222Lys		HCM	[26]
Gln222Arg		HCM	[38]
Ala223Thr		DCM	[51]
Ala223Val		LVNC	[7]

Asn224Ile		HCM	[47]
Ala226Thr		HCM	[52]
Leu227Val		HCM	[53]
Phe230Ser		LVNC	[18]
Asn232His		HCM	[3]
Asn232Ser		HCM	[54]
Ala233Ser		HCM	[55]
Thr235Asn		HCM	[19]
Val236Ala	Switch-1	HCM	[3]
Val236Ile	Switch-1	DCM	[56]
Arg237Gln	Switch-1	HCM	[3]
Arg237Trp	Switch-1	DCM	[57]
Asp239Asn	Switch-1	HCM	[43]
Asp239Glu	Switch-1	HCM	[24]
Ser241Phe	Switch-1	Ebstein anomaly	[58]
Arg243Cys	Switch-1	HCM	[59]
Arg243His	Switch-1	HCM	[60]
Phe244Leu	Switch-1	HCM	[26]
Phe244Cys	Switch-1	HCM	[3]
Gly245Glu	Switch-1	DCM	[61]
Lys246Gln		HCM	Rottbauer, 1996 HGMD
Lys246Ile		HCM	[3]
Phe247Leu		HCM	[1]
Ile248Thr		HCM	[5]
Ile248Phe		DCM	[61]
Arg249Gln		HCM	[62]
Arg249Gly		LVNC	[63]
Arg249Term		LVNC	[21]
Ile250Val		HCM	[3]
His251Asn		HCM	[64]
Phe252Cys		HCM	[65]
Phe252Leu		LVNC	[65]
Phe252Ser		HCM	[66]
Gly256Glu		HCM	[67]
Ala259Glu		HCM	[49]
Ile263Thr		HCM	[68]
Ile263Met		HCM	[23]
Thr265Asn		HCM	[24]
Thr265Ser		HCM	[3]
Tyr266Cys		HCM	[69]
Tyr266Term		LVNC	[18]
Leu267Val		HCM	[38]
Lys270Arg		HCM	[70]
Leu277Pro		HCM	[3]
Leu277Val		CM	[71]
Ala279Thr		DCM	[61]
Arg281Thr		LVNC	[72]
Tyr283Asp		Ebstein anomaly	[73]
Tyr283Cys		HCM	[5]
Tyr283His		LVNC	[74]
Ser291Phe		HCM	[75]

Leu301Gln		CM, non-compaction	[76]
Leu302Met		HCM	[6]
Ile303Met		HCM	[24]
Pro307Arg		HCM	[40]
Pro307His		HCM	[16]
Pro307Leu		HCM	[3]
Tyr308Cys		HCM	[25]
Tyr308Asn		HCM	[25]
Asp309Asn		HCM	[38]
Asp309Gly		HCM	Sabater-Molina, 2013 HGMD
Phe312Cys		HCM	[23]
Phe312Val		CM	[71]
Ile313Phe		HCM	[77]
Gln315Arg		LVNC	[19]
Glu317Gly		HCM	[16]
Thr318Asn		HCM	[24]
Thr318Pro		HCM	[77]
Val320Met		HCM	[78]
Ala321Val		HCM	[45]
Ser322Phe		HCM	[79]
Ser322Thr		HCM	[3]
Ile323Asn		HCM	[80]
Ile323Thr		HCM	[3]
Ala326Pro		HCM	[81]
Glu328Gly		HCM	[53]
Ala335Ser		HCM	[3]
Ala335Thr		HCM	[82]
Val338Met		HCM	[83]
Val338Ala		DCM	[56]
Phe341Leu		HCM	[6]
Glu344Lys		DCM	[84]
Glu345Asp		DCM	[31]
Lys346Glu		DCM	[31]
Met349Thr		HCM	[85]
Met349Val		DCM	[3]
Tyr350Asn		Ebstein anomaly	[73]
Lys351Glu		HCM	[28]
Glu354Ser		HCM	[5]
Ala355Ser		HCM	[3]
Ala355Thr		HCM	[10]
Ala355Val		HCM	[38]
Met362Arg	Loop 4	Ebstein anomaly, LVNC	[86]
Arg369Gln	Loop 4	CM, non-compaction	[87]
Glu374Val	Loop 4	HCM	[3]
Gly377Arg	Loop 4	DCM	[88]
Gly377Ser	Loop 4	DCM	[69]
Thr378Pro	Loop 4	HCM	[3]
Glu379Lys		HCM	[38]
Ala381Asp		HCM	[3]
Asp382Tyr		HCM	[64]
Lys383Arg		HCM	[3]

Lys383Asn		HCM	[89]
Ser384Pro		HCM	[19]
Ala385Val		HCM	[23]
Tyr386His		HCM	[50]
Tyr386Cys		DCM	[90]
Leu387Phe		CM	[91]
Met388Thr		HCM	[92]
Gly389Glu		HCM	[3]
Leu390Pro		Ebstein anomaly	[73]
Leu390Val		HCM	[78]
Asn391Thr		HCM	[13]
Ala393Val		HCM	[11]
Asp394Glu		HCM	[38]
Lys397Glu		LVNC	Mokhatar, 2017 HGMD
Gly398Glu		HCM	[70]
Pro402Ser	HCM Loop	HCM	[24]
Arg403Gln	HCM Loop	HCM	[93]
Arg403Gly	HCM Loop	HCM	[3]
Arg403Leu	HCM Loop	HCM	[94]
Arg403Pro	HCM Loop	LVNC	[18]
Arg403Trp	HCM Loop	HCM	[94]
Val404Leu	HCM Loop	HCM	[44]
Val404Met	HCM Loop	HCM	[23]
Val406Met	HCM Loop	HCM	[95]
Gly407Cys	HCM Loop	HCM	[96]
Gly407Val	HCM Loop	HCM	[23]
Asn408Lys	HCM Loop	HCM	[14]
Tyr410Asp	HCM Loop	HCM	[3]
Val411Ile	HCM Loop	HCM	[29]
Thr412Asn	HCM Loop	DCM	[46]
Gln418Lys		HCM	[47]
Tyr422Cys		CM	[97]
Ala423Val		HCM	[3]
Gly425Arg		HCM	[98]
Ala426Thr		HCM	[19]
Ala426Val		DCM	[84]
Leu427Met		HCM	[99]
Ala428Asp		LVNC	[100]
Ala428Val		HCM	[10]
Ala430Glu		HCM	[101]
Val431Leu		HCM	[24]
Arg434Thr		HCM	[19]
Met435Arg		HCM	[24]
Met435Thr		HCM	[53]
Asn437Asp		HCM	[13]
Trp438Term		MSM	[102]
Met439Arg		LVNC	[103]
Met439Leu		HCM	[3]
Met439Thr		HCM	[19]
Val440Ala		HCM	[14]
Val440Met		HCM and SM	[23]

Thr441Met		CM and distal myopathy	[104]
Arg442Cys		HCM	[25]
Arg442His		DCM	[105]
Ile443Thr		HCM	[10]
Ile443Val		HCM	[10]
Asn444Ser		HCM	[106]
Thr446Pro		HCM	[107]
Glu448Lys		LVNC	[7]
Thr449Asn		HCM	[82]
Thr449Ile		HCM	[3]
Thr449Ser		HCM	[3]
Lys450Glu		HCM	[108]
Lys450Thr		HCM	[98]
Gln451Pro		HCM	[3]
Arg453Cys		HCM	[109]
Arg453His		HCM	[53]
Arg453Ser		HCM	[110]
Arg453Leu		HCM	Haluza, 2000 HGMD
Arg453Pro		HCM	[82]
Ile457Arg		Myopathy with cardiac involvement	[111]
Ile457Thr		HCM	[17]
Asp461Glu		HCM	[19]
Ala463Asp	Switch 2	HCM	[82]
Glu466Gln	Switch 2	DCM	[84]
Ile467Leu	Switch 2	HCM	[112]
Ile467Thr	Switch 2	HCM	[17]
Phe468Leu	Switch 2	DCM	[90]
Asp469Asn	Switch 2	HCM	[24]
Asp469Tyr	Switch 2	DCM	[90]
Asn471Ser	Switch 2	HCM	[38]
Gln475His	Relay	DCM	[17]
Gln475Lys	Relay	DCM	[17]
Leu476Val	Relay	HCM	[3]
Ile478Asn	Relay	HCM	[6]
Asn479Thr	Relay	HCM	[11]
Asn479Ser	Relay	HCM	[10]
Glu483Cys	Relay	HCM	[25]
Glu483Lys	Relay	HCM	[113]
Phe489Ile	Relay	DCM	[17]
Met493Leu	Relay	HCM	[59]
Met493Ile	Relay	HCM	[114]
Met493Lys	Relay	HCM	[115]
Met493Val	Relay	HCM	[116]
Glu497Asp	Relay	HCM	[60]
Glu497Gly	Relay	HCM	[5]
Gln498Arg	Relay	HCM	[38]
Gln498Glu	Relay	LVNC	[117]
Glu499Gly	Relay	HCM	[3]
Glu499Lys	Relay	HCM	[118]
Glu500Ala	Relay	HCM	[28]

Tyr501Cys	Relay	HCM	[119]
GLy505Val		HCM	[3]
Ile506Phe		HCM	[25]
Ile506Thr		HCM	[19]
Ile511Phe		HCM	[98]
Ile511Thr		HCM	[23]
Phe513Cys		HCM	[120]
Gly514Asp		HCM	[3]
Met515Thr		HCM	[69]
Met515Val		HCM	[121]
Met515Arg		HCM	[23]
Asp516Glu		HCM	[122]
Leu517Met		HCM	[123]
Ile524Val	Activation loop	DCM	[61]
Glu525Lys	Activation loop	DCM	[90]
Pro527Leu	Activation loop	HCM	[40]
Pro527Thr	Activation loop	HCM	[3]
Gly529Asp	Activation loop	DCM	[17]
Ile530Val	Helix-loop-helix	HCM	[124]
Met531Arg	Helix-loop-helix	HCM	[125]
Ser532Pro	Helix-loop-helix	DCM	[105]
Ile533Val	Helix-loop-helix	DCM	[61]
Glu536Asp	Helix-loop-helix	HCM	[5]
Met539Ile	Helix-loop-helix	HCM	[126]
Met539Val	Helix-loop-helix	HCM	[127]
Met539Leu	Helix-loop-helix	HCM	[43]
Phe540Leu	Helix-loop-helix	DCM	[128]
Lys542Arg	Helix-loop-helix	HCM	[20]
Lys542Thr	Helix-loop-helix	LVNC	[129]
Ala543Thr	Helix-loop-helix	DCM	[17]
Thr544Ala	Helix-loop-helix	DCM	[3]
Asp545Asn	Helix-loop-helix	CM, non-compaction	[76]
Ala550Val	Helix-loop-helix	DCM	[46]
Asp554Glu	Helix-loop-helix	HCM	[25]
Asp554Tyr	Helix-loop-helix	HCM	[130]
Ala561Thr		HCM	[17]
Gln564Glu		DCM	[17]
Arg567His	Loop-3	DCM	[61]
Gly571Arg	Loop-3	HCM	[24]
His576Arg		HCM	[119]
His581Arg		HCM	[131]
Ala583Val		HCM	[1]
Gly584Arg		HCM	[109]
Gly584Ser		HCM	[29]
Ile585Phe		HCM	[3]
Val586Ala		HCM	[5]
Asp587Asn		HCM	[3]
Asp587Val		HCM	[132]
Ile590Val		HCM	[107]
Ile591Thr		DCM	[17]
Leu594Met		Myopathy	[133]

Asn597Lys		DCM	[61]
Leu601Val		HCM	[78]
Leu601Phe		HCM	[55]
Asn602Ser		HCM	[132]
Asn602Tyr		HCM	[3]
Glu603Lys		HCM	[24]
Val606Leu		HCM	[132]
Val606Met		HCM	[109]
GLy607Asp		HCM	[16]
Tyr609Cys		HCM	[17]
Ser612Pro		HCM	[25]
Lys615Gln		HCM	[98]
Lys615Asn		HCM	[134]
Thr619Ile		HCM	[3]
Leu620Pro		HCM	[135]
Tyr624Asn	Loop 2	HCM	[136]
Tyr624Cys	Loop 2	HCM	[16]
Pro630Ser	Loop 2	DCM	[3]
Ile631Thr	Loop 2	DCM	[3]
Lys635Term	Loop 2	DCM	[17]
Gly636Ser	Loop 2	HCM	[24]
Lys637Glu	Loop 2	DCM	[137]
Lys639Glu	Loop 2	LVNC	[18]
Ser642Leu	Loop 2	DCM	[51]
Ser648Leu		LVNC	[36]
Arg652Gly		HCM	[138]
Arg652Lys		HCM	[22]
Arg652Thr		HCM	[96]
Lys657Gln		HCM	[139]
Leu658Val		LVNC	[140]
Met659Ile		HCM	[10]
Met659Thr		DCM	[141]
Arg660Asn		HCM	[112]
Arg663Cys		HCM	[23]
Arg663Ser		HCM	[10]
Arg663His		HCM	[142]
His668Asn		HCM	[11]
Val670Ala		Myaglia	[143]
Arg671Cys		HCM	[10]
Arg671His		HCM	[19]
Cys672Phe		DCM	[3]
Glu677Val		LVNC	[7]
Met690Thr		HCM	[16]
Leu693Arg		LVNC	[7]
Arg694Cys		HCM	[29]
Arg694His		HCM	[144]
Arg694Leu		HCM	[145]
Asn696Ser		HCM	[146]
Val698Ala		HCM	[32]
Glu700Gly		LVNC	[36]
Gly701Asp		HCM	[147]

Gly701Ser		HCM	[3]
Ile702Asn		HCM	[5]
Ile702Val		HCM	[114]
Arg703His		HCM	[25]
Cys705Trp		HCM	[17]
Gly708Ala		HCM	[80]
Gly708Asp		HCM	[148]
Pro710Arg	Converter	HCM	[64]
Pro710His	Converter	HCM	[149]
Pro710Leu	Converter	HCM	[5]
Arg712His	Converter	LVNC	[7]
Arg712Leu	Converter	HCM	[150]
Gly716Arg	Converter	HCM	[120]
Gly716Ala	Converter	HCM	[38]
Asp717Gly	Converter	HCM	[151]
Arg719Trp	Converter	HCM	[120]
Arg719Gln	Converter	HCM	[152]
Arg719Leu	Converter	HCM	[14]
Arg719Pro	Converter	HCM	[1]
Gln720His	Converter	LVNC	[74]
Arg721Lys	Converter	HCM	[69]
Arg723His	Converter	HCM	[43]
Arg723Cys	Converter	HCM	[109]
Arg723Gly	Converter	HCM	[153]
Ile724Asn	Converter	DCM	[61]
Arg726Lys	Converter	HCM	[24]
Ala728Val	Converter	HCM	[154]
Ala729Pro	Converter	HCM	[4]
Ile730Asn	Converter	HCM	[155]
Ile730Met	Converter	HCM	[24]
Ile730Thr	Converter	HCM	[47]
Pro731Ala	Converter	HCM	[5]
Pro731Leu	Converter	HCM	[156]
Pro731Ser	Converter	HCM	[25]
Gly733Arg	Converter	DCM	[17]
Gly733Glu	Converter	HCM	[10]
Gly733Val	Converter	HCM	[25]
Gln734Glu	Converter	HCM	[123]
Gln734Pro	Converter	HCM	[98]
Ile736Leu	Converter	HCM	[50]
Ile736Val	Converter	HCM	[14]
Ile736Thr	Converter	HCM	[29]
Ile736Met	Converter	HCM	[8]
Ser738Asn	Converter	HCM	[157]
Ser738Thr	Converter	HCM	[24]
Arg739Ser	Converter	HCM	[5]
Lys740Asn	Converter	HCM	[19]
Gly741Ala	Converter	HCM	[23]
Gly741Arg	Converter	HCM	[67]
Gly741Trp	Converter	HCM	[8]
Ala742Glu	Converter	HCM	[158]

Glu743Asp	Converter	HCM	[159]
Ser748Pro	Converter	DCM	[160]
Leu749Gln	Converter	DCM	[56]
Phe758Cys	Converter	HCM	[3]
Thr761Asn	Converter	HCM	[161]
Lys762Arg	Converter	HCM	[5]
Val763Met	Converter	HCM	[162]
Val763Gly	Converter	HCM	[28]
Phe764Tyr	Converter	HCM	[114]
Phe764Leu	Converter	DCM	[105]
Lys766Gln	Converter	HCM	[5]
Gly768Arg	Converter	HCM	[43]
Leu769Pro	Converter	HCM	[24]
Glu774Val	Converter	HCM	[163]
Arg777Lys	Converter	DCM	[84]
Asp778Asn	Pliant	HCM	[97]
Asp778Gly	Pliant	HCM	[164]
Asp778Val	Pliant	HCM	[23]
Asp778Glu	Pliant	HCM	[122]
Glu779Asp	Pliant	HCM	[50]
Glu779Term	Pliant	HCM	[163]
Leu781Met	Pliant	HCM	[19]
Leu781Pro	Pliant	HCM	[5]
Ser782Arg	Pliant	HCM	[70]
Ser782Asn	Pliant	HCM	[26]
Arg783Cys	Pliant	HCM	[24]
Arg783His	Pliant	HCM	[25]
Arg783Pro	Pliant	CM and distal myopathy	[158]
Ile785Val	LCD	HCM	[3]
Arg787Cys	LCD	HCM	[162]
Arg787His	LCD	HCM	[10]
Arg793Gln	LCD	HCM	[17]
Leu796Phe	LCD	HCM	[29]
Ala797Pro	LCD	HCM	[92]
Ala797Thr	LCD	HCM	[165]
Tyr801Asn	LCD	HCM	[3]
Leu805Pro	LCD	DCM	[84]
Arg807Gly	LCD	LVNC	[27]
Arg807His	LCD	HCM	[77]
Asp809Tyr	LCD	HCM	[3]
Leu811Pro	LCD	HCM	[5]
Ile814Ser	LCD	DCM	[3]
Gln815Pro	LCD	HCM	[126]
Asn817Lys	LCD	HCM	[24]
Ile818Asn	LCD	CM, LVNC	[140]
Arg819Gln	LCD	HCM	[38]
Arg819Trp	LCD	DCM	[3]
Ala820Asp	LCD	HCM	[166]
Phe821Ser	LCD	HCM	[3]
Met822Leu	LCD	HCM	[45]
Met822Val	LCD	HCM	[1]

Met822Thr	LCD	HCM	[98]
Gly823Glu	LCD	HCM	[98]
Val824Ala	LCD	HCM	[5]
Val824Leu	LCD	HCM	[3]
Val824Ile	LCD	HCM	[29]
Trp827Cys	LCD	HCM	[50]
Pro828Ser	LCD	HCM	[20]
Tyr833Cys	LCD	HCM	[3]
Tyr833His	LCD	HCM	[24]
Phe834Leu	LCD	HCM	[5]
Phe834Tyr	LCD	HCM	[3]
Lys835Thr	LCD	HCM	[4]
Ile836Met	LCD	HCM	[24]
Ile836Thr	LCD	HCM	[147]
Pro838Leu	S2	HCM	[167]
Leu840Met	S2	HCM	[168]
Ser842Arg	S2	HCM	[169]
Ser842Asn	S2	HCM	[5]
Ser842Gly	S2	HCM	[24]
Glu844Lys	S2	HCM	[122]
Arg845Gly	S2	HCM	[170]
Glu846Gln	S2	HCM	[78]
Glu846Lys	S2	HCM	[24]
Lys847Glu	S2	HCM	[64]
Glu848Gly	S2	HCM	[5]
Met849Thr	S2	HCM	[171]
Ala850Asp	S2	HCM	[172]
Ala850Thr	S2	HCM	[4]
Met852Thr	S2	HCM	[10]
Lys853Gln	S2	HCM	[19]
Thr857Ile	S2	HCM	[3]
Thr857Pro	S2	HCM	[173]
Arg858Cys	S2	HCM	[23]
Arg858His	S2	HCM	[98]
Arg858Gly	S2	HCM	[83]
Arg858Pro	S2	HCM	[55]
Arg858Ser	S2	HCM	[131]
Lys860Glu	S2	HCM	[77]
Glu861Term	S2	DCM	[17]
Ala862Val	S2	HCM	[47]
Lys865Arg	S2	HCM	[55]
Lys865Glu	S2	HCM	[24]
Lys865Met	S2	HCM	[14]
Ser866Tyr	S2	HCM	[170]
Ser866Pro	S2	HCM	[45]
Ala868Pro	S2	HCM	[50]
Arg869Cys	S2	HCM	[120]
Arg869Gly	S2	HCM	[174]
Arg869His	S2	HCM	[43]
Arg869Leu	S2	HCM	[3]
Arg870Cys	S2	HCM	[175]

Arg870His	S2	HCM	[176]
Arg870Leu	S2	HCM	[177]
Leu873Pro	S2	HCM	[114]
Glu874Gln	S2	DCM	[178]
Glu874Val	S2	DCM	[84]
Met877Ile	S2	HCM	[3]
Met877Lys	S2	HCM	[95]
Val878Ala	S2	HCM	[179]
Val878Met	S2	HCM	[19]
Val878Gly	S2	HCM	[50]
Gln882Glu	S2	HCM	[28]
Asn885Lys	S2	HCM	[114]
Asn885Thr	S2	HCM	[180]
Gln888Lys	S2	HCM	[135]
Leu889His	S2	HCM	[64]
Gln890Pro	S2	HCM	[6]
Gln892Lys	S2	HCM	[13]
Ala893Val	S2	DCM	[90]
Glu894Gln	S2	HCM	[14]
Glu894Lys	S2	HCM	[19]
Glu894Gly	S2	HCM	[23]
Glu894Asp	S2	DCM	[3]
Gln895Lys	S2	HCM	[3]
Gln895Glu	S2	DCM	[3]
Leu898Val	S2	HCM	[5]
Asp900Glu	S2	HCM	[14]
Asp900Gly	S2	HCM	[3]
Asp900Val	S2	HCM	[24]
Ala901Gly	S2	HCM	[119]
Ala901Pro	S2	HCM	[92]
Glu902Lys	S2	CM restrictive	[24]
Glu903Lys	S2	HCM	[23]
Glu903Gly	S2	HCM	[162]
Glu903Gln	S2	HCM	[16]
Arg904Cys	S2	DCM	[181]
Arg904His	S2	DCM	[17]
Cys905Arg	S2	CM, non-compaction	[182]
Cys905Phe	S2	HCM	[29]
Cys905Tyr	S2	HCM	[19]
Asp906Asn	S2	HCM	[3]
Asp906Gly	S2	HCM	[138]
Gln907Lys	S2	HCM	[3]
Leu908Val	S2	HCM	[67]
Ile909Met	S2	HCM	[70]
Ile909Val	S2	HCM	[3]
Lys910Gln	S2	DCM	[17]
Lys912Asn	S2	HCM	[183]
Lys912Gln	S2	HCM	[183]
Ile913Met	S2	HCM	[24]
Ile913Thr	S2	HCM	[19]
Gln914His	S2	HCM	[5]

Leu915Pro	S2	LVNC	[36]
Glu916Lys	S2	HCM	[3]
Glu921Lys	S2	HCM	[23]
Glu924Gln	S2	HCM	[29]
Glu924Lys	S2	HCM	[109]
Arg925Gly	S2	DCM	[184]
Leu926Pro	S2	HCM	[126]
Leu926Val	S2	HCM	[3]
Glu927Lys	S2	HCM	[53]
Asp928Ala	S2	HCM	[3]
Asp928Asn	S2	HCM	[29]
Asp928Gly	S2	HCM	[6]
Asp928Val	S2	HCM	[185]
Glu929Lys	S2	CM, LVNC	[63]
Glu930Lys	S2	HCM	[26]
Glu930Gln	S2	HCM	[55]
Glu931Ala	S2	HCM	[157]
Glu931Gly	S2	HCM	[6]
Glu931Lys	S2	HCM	[23]
Met932Lys	S2	HCM	[43]
Glu935Lys	S2	HCM	[25]
Glu935Val	S2	HCM	[186]
Arg941Cys	S2	LVNC	[7]
Glu949Lys	S2	HCM	[109]
Glu949Val	S2	HCM	[24]
Asp953His	S2	HCM	[23]
Asp953Val	S2	HCM	[23]
Ile954Val	S2	DCM	[24]
Asp955Asn	S2	CM, non-compaction	[76]
Asp956Asn	S2	HCM	[97]
Leu957Arg	S2	HCM	[183]
Glu958Lys	S2	HCM	[97]
Leu959Arg	S2	HCM	[183]
Leu961Arg	S2	HCM	[112]
Leu961Pro	S2	LVNC	[36]
Leu961Val	S2	HCM	[3]
Ala962Asp	S2	HCM	[183]
Val964Leu	S2	DCM	[57]
Glu965Gly	S2	HCM	[3]
Glu965Lys	S2	HCM	[70]
Glu967Lys	S2	HCM	[5]
His969Pro	S2	HCM	[3]
Ala970Val	S2	DCM	[57]
Glu981Lys	S2	DCM	[48]
Met982Thr	S2	HCM	[25]
Lys991Asn	S2	DCM	[61]
Leu992Met	S2	HCM	[16]
Lys994Arg	S2	HCM	[19]
Ala1006Thr	S2	HCM	[157]
Lys1016Glu	S2	HCM	[3]
Thr1019Asn	S2	DCM	[46]

Lys1022Glu	S2	HCM	[3]
Leu1027Pro	S2	DCM	[17]
Leu1038Pro	S2	DCM	[137]
Val1044Ala	S2	HCM	[50]
Arg1045Cys	S2	HCM	[70]
Arg1045His	S2	HCM	[25]
Arg1045Leu	S2	HCM	[5]
Met1046Ile	S2	HCM	[3]
Arg1050Gln	S2	HCM	[19]
Ala1051Ala	S2	HCM	[106]
Ala1051Val	S2	DCM	[90]
Arg1053Gln	S2	DCM	[187]
Glu1056Asp	S2	HCM	[38]
Gly1057Asp	S2	HCM	[32]
Gly1057Ser	S2	HCM	[23]
Glu1070Lys	S2	HCM	[3]
Asp1077Glu	S2	HCM	[3]
Arg1079Gln	S2	HCM	[188]
Arg1079Gln	S2	Sudden unexpected death	[189]
Leu1090Val	S2	HCM	[13]
Asn1091Asp	S2	Sudden cardiac death	[190]
Glu1095Gly	S2	DCM	[3]
Asp1096Tyr	S2	DCM	[57]
Glu1097Lys	S2	HCM	[3]
Gly1101Ser	S2	HCM	[112]
Lys1109Glu	S2	HCM	[3]
Arg1114His	S2	HCM	[3]
Glu1116Lys	S2	HCM	[50]
Glu1119Lys	S2	HCM	[19]
Glu1120Lys	S2	HCM	[14]
Glu1123Gln	S2	HCM	[186]
Glu1125Gln	S2	HCM	[3]
Ala1128Thr	S2	HCM	[24]
Glu1133Lys	S2	HCM	[14]
Leu1135Arg	S2	HCM	[10]
Arg1136His	S2	HCM	[24]
Glu1142Lys	S2	HCM	[19]
Leu1143Arg	S2	HCM	[3]
Glu1152Val	S2	DCM	[61]
Gly1155Glu	S2	DCM	[3]
Thr1157Ala	S2	LVNC	Mokhtar, 2017 HGMD
Val1159Met	S2	HCM	[3]
Gln1160Arg	S2	HCM	[25]
Glu1162Lys	S2	HCM	[3]
Lys1165Glu	S2	HCM	[3]
Glu1168Asp	S2	HCM	[13]
Leu1183Gln	S2	HCM	[3]
Arg1193His	S2	DCM	[90]
Arg1193Ser	S2	DCM	[46]
His1196Tyr	S2	LVNC	[27]
Asp1198Gly	S2	HCM	[6]

Asp1198Tyr	S2	HCM	[77]
Ser1199Arg	S2	HCM	[24]
Gly1204Arg	S2	HCM	[24]
Glu1205Lys	S2	HCM	[25]
Asp1208Asn	S2	HCM	[25]
Asp1208Gly	S2	HCM	[191]
Asn1209Ser	S2	DCM	[56]
Val1213Met	S2	HCM	[24]
Gln1215His	S2	HCM	[114]
Lys1216Met	S2	HCM	[107]
Glu1218Gln	LMM	HCM	[10]
Ser1222Gly	LMM	HCM	[3]
Glu1223Gln	LMM	DCM	[17]
Glu1223Lys	LMM	DCM	[84]
Asn1234Thr	LMM	HCM	[24]
Ile1239Val	LMM	CM, right ventricular	[192]
Lys1242Lys	LMM	Sudden cardiac death	[193]
Leu1245Gln	LMM	DCM	[3]
Arg1250Pro	LMM	HCM	[194]
Arg1250Trp	LMM	CM, non-compaction	[87]
Ala1263Glu	LMM	HCM	[50]
Arg1268His	LMM	Cardiac defects	[195]
Arg1268Pro	LMM	HCM	[3]
Leu1273Pro	LMM	HCM	[25]
Arg1277Gln	LMM	HCM	[13]
Arg1277Pro	LMM	LVNC	[27]
Glu1286Lys	LMM	DCM	[90]
Glu1293Lys	LMM	HCM	[82]
Leu1297Gln	LMM	HCM	[19]
Leu1297Val	LMM	HCM	[50]
Gln1300Leu	LMM	HCM	[3]
Arg1303Gly	LMM	DCM	[141]
Arg1303Term	LMM	DCM	[31]
Asp1314Glu	LMM	DCM	[196]
Ala1325Val	LMM	DCM	[3]
Asn1327Lys	LMM	HCM	[197]
Ala1328Thr	LMM	DCM	[61]
Ala1332Thr	LMM	DCM	[17]
Gln1334Term	LMM	HCM	[197]
Ser1335Leu	LMM	DCM	[3]
Arg1337Gln	LMM	HCM	[25]
Arg1344Gln	LMM	HCM	[198]
Arg1344Trp	LMM	DCM	[90]
Gln1346Term	LMM	DCM	[149]
Tyr1347Cys	LMM	HCM	[147]
Glu1348Gln	LMM	HCM	[19]
Glu1348Lys	LMM	HCM	[19]
Glu1350Lys	LMM	DCM	[199]
Thr1351Met	LMM	HCM	[55]
Glu1356Gln	LMM	HCM	Sabatar-Molina, 2013 HGMD
Glu1356Lys	LMM	HCM	[23]

Arg1359Cys	LMM	CM, non-compaction	[65]
Val1360Ile	LMM	HCM	[19]
Ala1364Pro	LMM	LVNC	[74]
Gln1370Lys	LMM	HCM	[3]
Tyr1375His	LMM	HCM	[24]
Tyr1375Cys	LMM	HCM	[5]
Tyr1375Term	LMM	LVNC	[18]
Thr1377Met	LMM	HCM	[25]
Asp1378His	LMM	HCM	[3]
Ala1379Asp	LMM	HCM	[3]
Ala1379Thr	LMM	HCM	[200]
Arg1382Trp	LMM	HCM	[10]
Arg1382Gln	LMM	HCM	[99]
Leu1386Phe	LMM	HCM	[3]
Glu1387Lys	LMM	HCM	[24]
Ala1400Gly	LMM	HCM	[194]
Ala1403Asp	LMM	HCM	[24]
Val1404Met	LMM	HCM	[19]
Leu1414Met	LMM	HCM	[162]
Thr1417Ile	LMM	HCM	[6]
Arg1420Gln	LMM	HCM	[201]
Arg1420Leu	LMM	HCM	[3]
Arg1420Trp	LMM	HCM	[23]
Glu1426Lys	LMM	DCM	[46]
Leu1428Ser	LMM	HCM	[124]
Arg1434Cys	LMM	HCM/DCM	[17]
Ala1434Pro	LMM	Myopathy	[202]
Ser1435Pro	LMM	Myopathy	[203]
Ala1437Pro	LMM	Myopathy	[202]
Ala1439Pro	LMM	Myopathy	[204]
Asp1443Asn	LMM	HCM	[24]
Lys1444Glu	LMM	DCM	[56]
Asn1448Ser	LMM	HCM	[3]
Asp1450Asn	LMM	DCM	[90]
Leu1453Pro	LMM	Myopathy	[205]
Ala1454Thr	LMM	HCM	[119]
Glu1455Term	LMM	HCM	[188]
Glu1455Lys	LMM	HCM	[206]
Lys1459Asn	LMM	HCM	[23]
Ser1465Leu	LMM	DCM	[17]
Leu1467Val	LMM	Myopathy with cardiac involvement	[207]
Glu1468Gln	LMM	HCM	[24]
Glu1468Lys	LMM	HCM	[24]
Ser1470Pro	LMM	DCM	[61]
Gln1471Term	LMM	DCM	[61]
Glu1473Gly	LMM	HCM	[3]
Arg1475Cys	LMM	HCM	[25]
THr1479Ile	LMM	HCM	[3]
Leu1481Pro	LMM	Myopathy	[208]
Ala1487Thr	LMM	Myopathy	[209]

Tyr1488Cys	LMM	CM, non-compaction	[140]
Glu1489Gly	LMM	HCM	[25]
Ser1491Cys	LMM	HCM	[197]
Leu1492Pro	LMM	Myopathy	[133]
His1494Leu	LMM	HCM	[25]
Glu1496Ala	LMM	HCM	[210]
Arg1500Trp	LMM	DCM	[187]
Arg1500Pro	LMM	Myopathy	[211]
Gln1506Term	LMM	DCM	[17]
Ile1509Leu	LMM	HCM	[3]
Asp1511Ala	LMM	HCM	[3]
Thr1513Ser	LMM	HCM	[23]
Glu1514Lys	LMM	DCM	[56]
His1524Arg	LMM	HCM	[114]
Arg1530Term	LMM	CM	[212]
Glu1536Lys	LMM	DCM	[17]
Glu1541Pro	LMM	Myopathy	[208]
Glu1546Gln	LMM	HCM	[3]
Glu1546Asp	LMM	DCM	[3]
Glu1548Val	LMM	DCM	[3]
Ala1549Pro	LMM	Myopathy	[213]
Glu1554Gln	LMM	DCM	[214]
Glu1554Lys	LMM	HCM	[3]
Glu1555Gly	LMM	HCM	[99]
Glu1555Lys	LMM	HCM	[215]
Arg1560Gln	LMM	HCM	[24]
Arg1560Pro	LMM	Myopathy	[216]
Gln1567Term	LMM	Myopathy	[217]
Glu1573Lys	LMM	Ebstein anomaly	[73]
Arg1574Gln	LMM	HCM	[218]
Arg1574Trp	LMM	DCM	[61]
Arg1588Pro	LMM	Myopathy	[207]
Asn1589Lys	LMM	HCM	[19]
Leu1591Gln	LMM	Sudden cardiac death	[219]
Leu1591Pro	LMM	Myopathy	[220]
Ser1596Leu	LMM	HCM	[3]
Leu1597Arg	LMM	Myopathy	[221]
Thr1599Pro	LMM	Myopathy	[208]
Leu1601Pro	LMM	Myopathy	[222]
Ala1603Pro	LMM	Myopathy	[220]
Arg1606Cys	LMM	HCM	[80]
Arg1606His	LMM	HCM	[114]
Arg1608Pro	LMM	Myopathy, DCM	[223]
Arg1608Ser	LMM	Myopathy	[222]
Glu1610Gln	LMM	DCM	[224]
Ala1611Val	LMM	HCM	[225]
Leu1612Pro	LMM	Myopathy	[208]
Glu1619Lys	LMM	DCM	[57]
Leu1622Phe	LMM	HCM	[24]
Arg1634Cys	LMM	DCM	[46]
Ala1636Pro	LMM	Myopathy	[208]

Ala1637Thr	LMM	HCM	[50]
Glu1638Lys	LMM	HCM	[13]
Gln1640Arg	LMM	HCM	[3]
Leu1646Pro	LMM	Myopathy	[208]
Gln1647His	LMM	HCM	[13]
Asp1652Tyr	LMM	HCM	[106]
Ala1660Glu	LMM	DCM	[17]
Arg1662His	LMM	DCM	[17]
Ala1663Pro	LMM	Myopathy	[211]
Asn1664Lys	LMM	HCM	[24]
Lys1668Glu	LMM	HCM	[3]
Val1674Met	LMM	HCM	[3]
Asn1674Leu	LMM	DCM	[3]
Arg1676Pro	LMM	Myopathy	[226]
Arg1677Cys	LMM	HCM	[3]
Arg1677His	LMM	DCM	[17]
Val1691Met	LMM	HCM	[10]
Glu1696Asp	LMM	HCM	[3]
Arg1697Trp	LMM	DCM	[31]
Gln1704Term	LMM	HCM	[3]
Leu1706Pro	LMM	Myopathy	[211]
Leu1706Val	LMM	CM	[227]
Ile1707Phe	LMM	HCM	[24]
Arg1712Trp	LMM	HCM	[197]
Arg1712Gln	LMM	HCM	[13]
His1717Gln	LMM	HCM	[228]
Gln1719Arg	LMM	HCM	[24]
Leu1723Pro	LMM	Myopathy	[229]
Ile1724Met	LMM	HCM	[3]
Lys1727Term	LMM	DCM	[31]
Asp1731Val	LMM	DCM	[56]
Ala1744Ser	LMM	Sudden cardiac death	[230]
Cys1748Tyr	LMM	Sudden cardiac death	[190]
Glu1752Lys	LMM	HCM	[231]
Glu1752Val	LMM	HCM	[17]
Glu1753Lys	LMM	HCM	[197]
Lys1757Glu	LMM	HCM	[19]
Thr1760Arg	LMM	HCM	[191]
Thr1760Met	LMM	HCM	[172]
Ala1763Thr	LMM	HCM	[25]
Met1764Lys	LMM	HCM	[3]
Met1765Lys	LMM	HCM	[6]
Ala1766Thr	LMM	CM	[65]
Glu1768Lys	LMM	HCM	[23]
Leu1769Met	LMM	HCM	[55]
Gln1773Lys	LMM	HCM	[194]
Thr1775Ile	LMM	HCM	[14]
Ser1776Gly	LMM	HCM	[197]
Ala1777Thr	LMM	HCM	[10]
His1778Tyr	LMM	HCM	[3]
Leu1779Pro	LMM	Myopathy	[208]

Arg1781Cys	LMM	HCM	[25]
Arg1781His	LMM	HCM	[6]
Met1782Val	LMM	HCM	[124]
Asp1792Gly	LMM	DCM	[17]
Leu1793Pro	LMM	Myopathy	[232]
Gln1794Lys	LMM	HCM	[233]
Gln1794Glu	LMM	DCM	[90]
Leu1797Pro	LMM	DCM	[192]
GLu1799Lys	LMM	LVNC	[74]
Glu1801Gly	LMM	DCM	[17]
Glu1801Lys	LMM	Myopathy/DCM	[61, 220]
Ala1804Thr	LMM	DCM	[234]
Gly1808Ala	LMM	DCM	[57]
Gly1808Ser	LMM	HCM	[114]
Arg1818Trp	LMM	HCM	[25]
Arg1820Gln	LMM	CM, distal myopathy	[235]
Arg1820Trp	LMM	Myopathy	[236]
Asn1824Cys	LMM	HCM	[3]
Asn1824Ser	LMM	HCM	[17]
Glu1829Gly	LMM	HCM	[20]
Arg1832Cys	LMM	DCM	[137]
Ala1834Thr	LMM	HCM	[24]
Glu1835Term	LMM	HCM	[19]
Ser1836Leu	LMM	HCM	[137]
Lys1838Glu	LMM	DCM	[84]
Ser1843Gly	LMM	HCM	[24]
Glu1844Lys	LMM	DCM	[3]
Arg1845Trp	LMM	Myopathy	[237]
Arg1846Cys	LMM	HCM	[50]
Lys1848Thr	LMM	HCM	[13]
Tyr1852Term	LMM	DCM	[84]
Thr1854Met	LMM	HCM	[23]
Thr1854Thr	LMM	DCM	[238]
Glu1856Lys	LMM	LVNC	[220]
Arg1858Met	LMM	HCM	[47]
Arg1863Trp	LMM	HCM	[3]
Arg1863Gln	LMM	DCM	[57]
Asp1869Gly	LMM	HCM	[24]
Val1875Phe	LMM	DCM	[196]
Lys1879Glu	LMM	DCM	[169]
Arg1880His	LMM	HCM	[157]
Glu1883Lys	LMM	Myopathy, HCM	[239]
Glu1886Lys	LMM	Myopathy	[220]
Glu1887Gly	LMM	Sudden cardiac death	[240]
Arg1897His	LMM	HCM	[6]
Val1899Ala	LMM	HCM	[24]
His1901Gln	LMM	DCM	[57]
His1901Leu	LMM	Myopathy	[241]
Glu1902Gln	LMM	HCM	[33]
Glu1902Lys	LMM	DCM	[56]
Ala1906Gly	LMM	DCM	[84]

Arg1909Pro	LMM	DCM	[212]
Arg1909Trp	LMM	HCM	[3]
Glu1914Lys	LMM	DCM	[90]
Val1917Phe	LMM	HCM	[3]
Asn1918Lys	LMM	Ebstein anomaly	[73]
Arg1925Cys	LMM	HCM	[3]
Arg1925Gly	LMM	LVNC	[140]
Ile1927Phe	LMM	HCM	[50]
Thr1929Met	LMM	HCM	[23]
Gly1931Cys	LMM	HCM	[99]
Thr1929Met	LMM	HCM	[23]
Term1936Leu		Myopathy	[242]
Term1936Trp		Myopathy	[243]
Term1936Tyr		Myopathy	[133]

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Supplemental Table 2: Missense mutations in MYH6 (α-cardiac myosin heavy chain) and their associated disease. HCM: hypertrophic cardiomyopathy. DCM: dilated cardiomyopathy. LVNC: Left Ventricular non-compaction. SUD: sudden unexplained death

Mutations	Myosin region	Disease	Reference
Ala12Val		HCM	[1]
Arg17Cys		DCM	[2]
Arg17His		Atrial septal defect	[3]
Arg17Leu		LVNC	[4]
Arg23His		HCM	[5]
Pro40Arg	SH3-like fold	Ventricular septal defect	[6]
Arg54Gln	SH3-like fold	long QY syndrome	[7]
Gly56Arg	SH3-like fold	HCM	[8]
Val71Ala	SH3-like fold	DCM	[9]
Glu98Lys		DCM	[10]
Val101Met		DCM	[9]
Arg108Cys		Congenital heart disease	[11]
Ala110Thr		DCM	[12]
Tyr115Asn		Hypoplastic left heart syndrome	[13]
Tyr162Cys		DCM	[12]
Ala199Ser	Loop-1	DCM	[9]
Arg204His	Loop-1	HCM	[14]
Asp208Asn	Loop-1	Atrioventricular septum defect	[15]
Ala230Pro		Congenital heart defects	[16]
Arg244His	Switch-1	Pulmonary hypertension	[17]
His252Gln		Congenital heart defects	[16]
Ile275Asn		DCM	[18]
Gln277His		Hypoplastic left heart syndrome	[13]
Pro308Leu		Pulmonary arterial hypertension	[17]
Ala336Gly		DCM	[10]
Met363Leu	Loop 4	HCM	[19]
Arg370Leu	Loop 4	LVNC	[20]
Gln373Pro	Loop 4	Congenital heart disease	[11]
Asp383Asn		Hypoplastic left heart syndrome	[13]
Ser385Leu		Hypoplastic left heart syndrome	[13]
Leu388Phe		Atrioventricular canal defects	[21]
Asp395GLu		SUD	[22]
Met436Val		Hypoplastic left heart syndrome	[13]
Arg443Cys		DCM	[10]
Arg443Pro		Hypoplastic left heart syndrome	[13]
His493Arg	Relay	SUD	[23]

Met494Ile	Relay	Left-ventricular outflow tract obstructions	[24]
Glu501Term		Congenital heart defects	[16]
Ile512Thr		Left ventricular obstruction with Shone complex	[21]
Glu526Lys	Activation loop	Pulmonary arterial hypertension	[17]
Cys539Arg	Helix-loop-helix	Atrial septal defect	[3]
Lys543Arg	Helix-loop-helix	Atrial septal defect	[3]
Arg568Cys	Loop-3	DCM	[18]
Gly585Cys		Pulmonary arterial hypertension	[17]
Gly585Ser		Left ventricular obstruction with Shone complex	[21]
Asp588Ala		Hypoplastic left heart syndrome	[25]
Asn678Ser		Congenital heart defects	[26]
Val700Met		Congenital heart defects	[16]
Ile704Asn		Hypoplastic left heart syndrome	[25]
Arg721Trp	Converter	Sick sinus syndrome	[27]
Ala769Val	Converter	Arrhythmogenic right ventricular cardiomyopathy	[28]
Arg795Gln	LCD	HCM	[29]
Arg795Trp	LCD	Hypoplastic left heart syndrome	[13]
Arg809Cys	LCD	HCM	[14]
Ile820Asn	LCD	Atrial septal defects	[30]
Pro830Leu	LCD	DCM	[31]
GLu846Asp	S2	DCM	[9]
Ala852Thr	S2	SUD	[22]
Arg860Cys	S2	Restrictive CM	[32]
Arg871His	S2	Cardiac arrest	[33]
Arg872Cys	S2	DCM	[10]
Ala936Val	S2	Atrioventricular septum defect	[15]
Lys942Term	S2	SUD young	[34]
Cys949Term	S2	Arrhythmogenic CM	[35]
Glu951Term	S2	Atrial septal defect	[23]
Asp955Val	S2	Bipolar	[36]
Ala964Ser	S2	Hypoplastic left heart syndrome	[13]
Glu1003Asp	S2	LVNC	[20]
Ala1004Ser	S2	DCM	[31]
Arg1047Cys	S2	DCM	[37]
Gln1065His	S2	HCM	[31]
Ile1068Thr	S2	Left ventricular obstruction with Shone complex	[21]
Lys1083Asn	S2	HCM	[32]
Ala1101Val	S2	Heart rate association with	[38]
Arg1116Cys	S2	DCM	[10]
Arg116Ser	S2	Congenital heart defects	[16]
Arg1143Gln	S2	DCM	[39]

Ser1149Arg	S2	DCM	[9]
Arg1151Gln	S2	Hypoplastic left heart syndrome	[13]
Arg1177Trp	S2	DCM	[18]
Glu1207Lys	S2	Hypoplastic left heart syndrome	[25]
Thr1253Met	LMM	DCM	[37]
Arg1279Term	LMM	Congenital heart defects	[40]
Glu1295Gln	LMM	Atrioventricular septum defect	[15]
Ala1298Val	LMM	Hypoplastic left heart syndrome	[13]
Ala1327Val	LMM	Atrioventricular canal defects	[21]
His1333Asn	LMM	Sudden unexplained death	[22]
Arg1339Trp	LMM	DCM	[41]
Ala1366Asp	LMM	Congenital heart defects	[16]
Thr1379Met	LMM	Hypoplastic left heart syndrome	[25]
Glu1389Lys	LMM	Sudden unexplained death	[22]
Arg1398Gln	LMM	Cardiac dysrhythmia	[42]
Val1406Met	LMM	DCM	[9]
Arg1422Leu	LMM	Pulmonary arterial hypertension	[17]
Asn1438Ile	LMM	Sudden unexplained death	[22]
Ala1440Pro	LMM	DCM	[18]
Ala1442Val	LMM	Abnormality of the mitochondrion	[43]
Ala1443Asp	LMM	Congenital heart defects	[16]
Glu1457Lys	LMM	DCM	[31]
Arg1502Gln	LMM	DCM	[18]
Glu1503Val	LMM	Hypoplastic left heart syndrome	[13]
Gln1543Lys	LMM	Sudden unexplained death	[22]
Phe1567Cys	LMM	DCM	[9]
Arg1576Gln	LMM	HCM	[44]
Glu1584Lys	LMM	Hypoplastic left heart syndrome	[13]
Arg1590Ser	LMM	Cardiac defects	[45]
Asn1591Ser	LMM	HCM	[46]
Arg1608Cys	LMM	Ventricular septal defect	[6]
Arg1610Cys	LMM	Atrial and ventricular septal defects	[21]
Lys1617Thr	LMM	LVNC	[20]
Glu1677GLn	LMM	Pulmonary arterial hypertension	[17]
Leu1690Arg	LMM	DCM	[41]
Arg1691Cys	LMM	HCM	[32]
Arg1699Trp	LMM	DCM	[39]
Ala1704Val	LMM	HCM	[47]
Ser1734Leu	LMM	Sudden unexplained death	[48]
Glu1754Term	LMM	Hypoplastic left heart syndrome	[13]
Met1766Val	LMM	DCM	[12]
Arg1820Gln	LMM	HCM	[49]

GLy1826Asp	LMM	Atrial septal defect	[6]
Lys1840Arg	LMM	Hypoplastic left heart syndrome	[13]
Glu1846Lys	LMM	HCM	[49]
Arg1865Gln	LMM	Congenital heart defects	[16]
Glu1885Lys	LMM	Wolff-parkinson-White syndrome	[50]
Ala1891Thr	LMM	Left ventricular obstruction with Shone complex	[21]
Arg1899Cys	LMM	DCM	[10]
Arg1899His	LMM	Left ventricular obstruction with Shone complex	[21]
Lys1932Term	LMM	Left ventricular obstruction with Shone complex	[21]

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Supplemental Table 3: Missense mutations in MYH3 (embryonic myosin heavy chain) and their associated disease. SCT: spondylocarpotarsal synostosis syndrome

Val40Met	Arthrogryposis	[1]
Tyr47Term	SCT	[2]
Thr178Ile	Arthrogryposis, distal, type 2A	[3]
Gly184Ala	Arthrogryposis, distal, type 1	[4]
Ala234Thr	Arthrogryposis, distal 2B & myosin myopathy	[5]
Gly246Ala	Distal arthrogryposis syndrome 2b	[4]
Ser261Phe	Arthrogryposis, distal, type 2B	[3]
Phe287Val	Sheldon-Hall syndrome & vertebral fusions	[6]
Ser292Cys	Arthrogryposis, distal, type 2B	[3]
Thr333Arg	SCT	[7]
Leu340Gln	Distal arthrogryposis syndrome 2b	[4]
Glu375Lys	Arthrogryposis, distal, type 2B	[3]
Tyr387Cys	Arthrogryposis, distal, type 2A	[8]
Phe437Ile	Arthrogryposis, distal, type 1	[9]
Asp462Gly	Arthrogryposis, distal 2B & myosin myopathy	[5]
Phe466Cys	Distal arthrogryposis syndrome 2b	[4]
Glu498Gly	Arthrogryposis, distal, type 2A	[3]
Lys504Asn	Arthrogryposis, distal, type 1	[4]
Asp517Tyr	Arthrogryposis, distal, type 2B	[3]
Tyr583Ser	Arthrogryposis, distal, type 2A	[3]
Asp629Asn	Autism spectrum disorder	[10]
Phe645Cys	SCT	[11]
Arg672Cys	Arthrogryposis, distal, type 2A	[3]
Arg672His	Arthrogryposis, distal, type 2A	[3]
Gly769Val	Arthrogryposis, distal, type 2B	[3]
Val825Asp	Arthrogryposis, distal, type 2A	[3]
Lys838Glu	Arthrogryposis, distal, type 2B	[3]
Gln1075Pro	Pterygium syndrome	[12]
Arg1290His	Cleft lip and/or palate	[13]
Leu1320Pro	Arthrogryposis multiplex congenita with axoglial defects	[14]
Leu1344Pro	SCT	[7]
Asp1622Ala	Arthrogryposis, distal, type 2B	[3]
Ala1637Val	Arthrogryposis, distal, type 2B	[3]
Lys1652Arg	Developmental disorder	[15]

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Supplemental Table 4: Missense mutations in MYH2 (MyHC-2a) and their associated disease.

Thr178Ile	Myopathy with external ophthalmoplegia	[1]
Ala236Thr	Myopathy with external ophthalmoplegia	[1]
Arg246His	Myopathy, MYH2-related	[2]
Ala387Val	Myopathy, proximal with ophthalmoplegia	[3]
Val423Met	Proximal myopathy	[4]
Arg445Cys	Myopathy with external ophthalmoplegia	[1]
Glu500Term	Myopathy with external ophthalmoplegia	[5]
Phe516Val	Congenital myopathy	[6]
Met531Thr	Myopathy with external ophthalmoplegia	[1]
His672Tyr	Pulmonary arterial hypertension	[7]
Glu706Lys	Inclusion body myopathy	[8]
Asp756Asn	Myopathy	[9]
Arg783Term	Myopathy, early onset	[10]
Leu802Term	Myopathy, early onset	[10]
Val805Ala	Inclusion body myositis	[11]
Lys890Ile	Pulmonary arterial hypertension	[7]
Val970Ile	Inclusion body myopathy	[12]
Ser1043Ala	Inclusion body myositis	[13]
Leu1061Val	Inclusion body myopathy	[12]
Gln1111Term	Congenital myopathy	[6]
Arg1218Term	Autism spectrum disorder	[14]
Lys1311Gln	Pulmonary arterial hypertension	[7]
Leu1420Phe	Neuromuscular disorder	[15]
Glu1681Lys	Inclusion body myositis	[13]
Leu1870Pro	Myopathy, MYH2-related	[16]
Leu1877Pro	Myopathy, distal and proximal with complete ophthalmoplegia	[17]

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