

Gene	Nucleotide change	Protein change	Variant type	No. of pts.	Mutation classification (as reported on original report)	ClinVar (n)	Ref	ExAC MAF (Allele Count)	UCSC Cons	UCSC Status	Grantham Score	PolyPhen 2	Condel	Mutation Assessor	SIFT
ACTC1	c.309C>A	p.His103Gln	Missense	1	Dominant, possibly associated	-		-	0	Pathogenic	Benign (24)	Possibly damaging	Deleterious	Medium	Tolerated
LAMP2	c.1171G>A	p.Val391Ile	Missense	1*	VUS	-		-	41	Benign	Benign (30)	-	Neutral	Neutral	Tolerated
LAMP2	c.755T>G	p.Ile252Ser	Missense	1	VUS	Benign		-	17	Benign	Pathogenic (142)	Possibly damaging	Neutral	Medium	Tolerated
MYBPC3	c.41A>G	p.Lys14Arg	Missense	1*	VUS	-		0.00121% (1/82480)	0	Pathogenic	Benign (26)	Possibly damaging	Neutral	Low	Tolerated
MYBPC3	c.177_187 del11	p.Glu60fs	Frameshift	1	Probable deleterious mutation	-		-	-	-	-	-	-	-	-
MYBPC3	c.436dupA	p.Thr146fs	Frameshift	1	Disease- causing mutation	-		-	-	-	-	-	-	-	-
MYBPC3	c.481C>A	p.Pro161Thr	Missense	1	Likely disease-causing mutation	Pathogenic (1), Uncertain (1)		0.00415% (1/24106)	0	Pathogenic	Benign (38)	Probably damaging	Deleterious	Medium	Damaging
MYBPC3	c.484C>T	p.Gln162Ter	Nonsense	1	Probable deleterious mutation	-		-	-	-	-	-	-	-	-
MYBPC3	c.506-17C>T	p.IVS4-17C>T	Splicing	1	Unknown**	Benign (1)		0.017845% (8/44830)	-	-	-	-	-	-	-
MYBPC3	c.655G>T	p.Val219Phe	Missense	1*	Deleterious mutation	Pathogenic (1)	13, 23	0.00106% (1/94000)	4	Benign	Benign (50)	Probably damaging	Deleterious	Medium	Damaging
MYBPC3	c.655G>C	p.Val219Leu	Missense	2	VUS	Pathogenic (2)		-	4	Benign	Benign (32)	Possibly damaging	Deleterious	Medium	Tolerated
MYBPC3	c.682G>A	p.Asp228Asn	Missense	1	VUS	Uncertain (1)	29, 33	0.00569% (6/105368)	12	Benign	Benign (23)	Benign	Neutral	Neutral	Damaging
MYBPC3	c.772G>A	p.Glu258Lys	Missense	4	Deleterious mutation	Pathogenic (6)	24, 34	0.00390% (3/76868)	1	Benign	Benign (57)	Possibly damaging	Deleterious	Medium	Damaging
MYBPC3	c.821+1G>A	p.IVS7+1G>A	Splicing	2	Probable deleterious mutation	-	23, 24	0.00431% (1/23204)	-	-	-	-	-	-	-
MYBPC3	c.927-9G>A	p.IVS11-9G>A	Splicing	2*	Disease- causing mutation	-		-	-	-	-	-	-	-	-
MYBPC3	c.927-2A>G	p.IVS11-2A>G	Splicing	1	Predicted deleterious mutation	-	24, 35	-	-	-	-	-	-	-	-
MYBPC3	c.1090+1G>T	p.IVS12+1G>T	Splicing	1	Dominant, probably associated	-		-	-	-	-	-	-	-	-
MYBPC3	c.1168delC	p.His390fs	Frameshift	2	Dominant, probably associated	-	13, 23	-	-	-	-	-	-	-	-
MYBPC3	c.1474delG	p.Glu492fs	Frameshift	1	Dominant, probably associated	-		-	-	-	-	-	-	-	-
MYBPC3	c.1813G>A	p.Asp605Asn	Missense	1	Published, disease-causing mutation**	Pathogenic (1) Uncertain (2)	23	0.01889% (9/47656)	2	Benign	Benign (23)	Benign	Neutral	Low	Tolerated
MYBPC3	c.1928-2A>G	p.IVS20-2A>G	Splicing	1	Published, disease-causing mutation	-	13, 10	-	-	-	-	-	-	-	-
MYBPC3	c.2096delC	p.Pro699fs	Frameshift	5	Predicted deleterious mutation	-	23, 25, 26	-	-	-	-	-	-	-	-
MYBPC3	c.2132G>A	p.Trp711Ter	Nonsense	1	Predicted deleterious mutation	-		-	-	-	-	-	-	-	-
MYBPC3	c.2308+1G>A	p.IVS23+1G>A	Splicing	1	Disease-causing mutation	-		-	-	-	-	-	-	-	-
MYBPC3	c.2373dupG	p.Trp792fs	Frameshift	2*	Deleterious mutation	Pathogenic (6)	27, 28	0.00378% (1/26444)	-	-	-	-	-	-	-

MYBPC3	c.2374T>C	p.Trp792Arg	Missense	2*	Published, disease-causing mutation	Pathogenic (4)	23	-	1	Benign	Pathogenic (101)	Probably damaging	Deleterious	High	Damaging
MYBPC3	c.2450G>A	p.Arg817Gln	Missense	1	Possible deleterious mutation	Uncertain (2)		0.00166% (2/120688)	0	Pathogenic	Benign (43)	Probably damaging	Deleterious	Medium	Damaging
MYBPC3	c.2497G>A	p.Ala833Thr	Missense	1	VUS**	Pathogenic (5), uncertain (2), Benign (2)	23, 29	0.16825% (203/120654)	1	Benign	Benign (58)	Possibly damaging	Neutral	Medium	Damaging
MYBPC3	c.2556delC	p.Ala851fs	Frameshift	1	Probable deleterious mutation	-	23	-	-	-	-	-	-	-	-
MYBPC3	c.2558delG	p.Gly853fs	Frameshift	1	Predicted deleterious mutation	-		0.00085% (1/117708)	-	-	-	-	-	-	-
MYBPC3	c.2614G>A	p.Glu872Lys	Missense	1	VUS**	Uncertain (2)		0.05867% (38/64766)	0	Pathogenic	Benign (57)	Probably damaging	Neutral	Medium	Tolerated
MYBPC3	c.2686G>A	p.Val896Met	Missense	1*	VUS**			1.27512% (297/23292)	17	Benign	Benign (22)	Benign	Neutral	Neutral	Damaging
MYBPC3	c.2728C>A	p.Pro910Thr	Missense	1	VUS**	Uncertain (4)		0.03906% (6/15362)	33	Benign	Benign (38)	Benign	Neutral	Neutral	Tolerated
MYBPC3	c.2864_2865 delCT	p.Pro955fs	Frameshift	5	Predicted deleterious mutation	Pathogenic (2)	23, 24	-	-	-	-	-	-	-	-
MYBPC3	c.2905C>T	p.Gln969Ter	Nonsense	1	Predicted deleterious mutation	Pathogenic (3)	23, 30	-	-	-	-	-	-	-	-
MYBPC3	c.3083C>T	p.Thr1028Ile	Missense	2	VUS	Pathogenic (1), Uncertain (1)		-	2	Benign	Benign (89)	Probably damaging	Deleterious	Medium	Damaging
MYBPC3	c.3107G>A	p.Arg1036His	Missense	1	VUS	Uncertain (1)		0.00581% (7/120414)	15	Benign	Benign (29)	Benign	Neutral	Low	Tolerated
MYBPC3	c.3124_3125 insAA	p.Thr1042fs	Frameshift	1	Published, disease-causing mutation	-	23, 24	-	-	-	-	-	-	-	-
MYBPC3	c.3190+2T>G	p.IVS29+2T>G	Splicing	1*	Presumed pathogenic			0.00169% (2/118438)	-	-	-	-	-	-	-
MYBPC3	c.3190+3delG	p.IVS29+3delG	Splicing	1	Disease-causing mutation	-		-	-	-	-	-	-	-	-
MYBPC3	c.3258G>A	p.Trp1086Ter	Nonsense	1	Predicted deleterious mutation	-		0.00218% (1/45884)	-	-	-	-	-	-	-
MYBPC3	c.3330+2T>G	p.IVS30+2T>G	Splicing	5	Published, disease-causing mutation	-	31, 32	-	-	-	-	-	-	-	-
MYBPC3	c.3407_3409 delACT	p.Tyr1136Del	InDel	1	VUS	Uncertain (1)		-	-	-	-	-	-	-	-
MYBPC3	c.3512delA	p.Asn1171fs	Frameshift	1	Dominant, probably associated	-		-	-	-	-	-	-	-	-
MYBPC3	c.3535G>A	p.Glu1179Lys	Missense	1*	VUS**			0.04592% (49/106716)	1	Benign	Benign (57)	Possibly damaging	Deleterious	Medium	Tolerated
MYBPC3	c.3624delC	p.Lys1209fs	Frameshift	1	Probable deleterious mutation	-		-	-	-	-	-	-	-	-
MYH7	c.427C>T	p.Arg143Trp	Missense	1	Disease-causing mutation	Pathogenic (3), Uncertain (1)		0.00494% (6/121402)	0	Pathogenic	Pathogenic (101)	Probably damaging	Deleterious	Medium	Damaging
MYH7	c.428G>A	p.Arg143Gln	Missense	1	VUS	-	44	0.00082%	0	Pathogenic	Benign (43)	Probably	Deleterious	Low	Damaging

								(1/121400)				damaging			
MYH7	c.715G>A	p.Asp239Asn	Missense	1	VUS	Pathogenic (2), Uncertain (1)	25	-	0	Pathogenic	Benign (23)	Probably damaging	Deleterious	Low	Tolerated
MYH7	c.746G>A	p.Arg249Gln	Missense	1	Deleterious mutation	Pathogenic (5)	45, 46	-	0	Pathogenic	Benign (43)	Probably damaging	Deleterious	Medium	Damaging
MYH7	c.1370T>C	p.Ile457Thr	Missense	1	VUS	Pathogenic (1), Uncertain (1)	36	0.00082% (1/121396)	0	Pathogenic	Benign (89)	Probably damaging	Deleterious	High	Damaging
MYH7	c.1588A>G	p.Ile530Val	Missense	1	Dominant, possibly associated	-		0.00082% (1/121380)	0	Pathogenic	Benign (30)	Benign	Deleterious	Medium	Damaging
MYH7	c.1750G>A	p.Gly584Ser	Missense	1	VUS	Pathogenic (2)	13	-	0	Pathogenic	Benign (55)	Benign	Deleterious	High	Damaging
MYH7	c.1988G>A	p.Arg663His	Missense	1	Deleterious mutation	Pathogenic (8)		0.00165% (2/121338)	2	Benign	Benign (29)	Possibly damaging	Deleterious	Low	Tolerated
MYH7	c.2206A>G	p.Ile736Val	Missense	1	VUS	Pathogenic (1), Uncertain (2)		0.00082% (1/121406)	1	Benign	Benign (30)	Probably damaging	Neutral	Neutral	Tolerated
MYH7	c.2221G>T	p.Gly741Trp	Missense	2	VUS	Pathogenic (4), Uncertain (1)	37, 38	-	2	Benign	Pathogenic (184)	Probably damaging	Deleterious	Medium	Damaging
MYH7	c.2346C>A	p.Ser782Arg	Missense	1	Possible deleterious mutation	Pathogenic (1)		-	4	Benign	Pathogenic (109)	Possibly damaging	Deleterious	Medium	Damaging
MYH7	c.2546T>C	p.Met849Thr	Missense	1	Possible deleterious mutation	Pathogenic (2)		-	4	Benign	Benign (81)	Probably damaging	Deleterious	Medium	Damaging
MYH7	c.2681A>G	p.Glu894Gly	Missense	1	VUS	Pathogenic (4), Uncertain (1)		-	1	Benign	Benign (98)	Probably damaging	Deleterious	Medium	Damaging
MYH7	c.2717A>G	p.Asp906Gly	Missense	1	Pathogenic	Pathogenic (4)	39	-	6	Benign	Benign (94)	Benign	Deleterious	Medium	Damaging
MYH7	c.2722C>G	p.Leu908Val	Missense	4	Deleterious mutation	Pathogenic (5)	40, 41	-	0	Pathogenic	Benign (32)	Probably damaging	Deleterious	High	Damaging
MYH7	c.2846A>T	p.Glu949Val	Missense	1	VUS	Pathogenic (2)		0.00082% (1/121412)	0	Pathogenic	Pathogenic (121)	Probably damaging	Deleterious	Medium	Damaging
MYH7	c.2945T>C	p.Met982Thr	Missense	1	VUS**	Pathogenic (1), Uncertain (4), Benign (1)	10, 42	0.09060% (110/121412)	0	Pathogenic	Benign (81)	Probably damaging	Deleterious	Medium	Damaging
MYH7	c.3066G>T	p.Lys1022Asn	Missense	1	VUS	-		-	0	Pathogenic	Benign (94)	Probably damaging	Deleterious	High	Damaging
MYH7	c.3134G>A	p.Arg1045His	Missense	1	VUS	-	43	0.00330% (1/121396)	0	Pathogenic	Benign (29)	Probably damaging	Deleterious	High	Damaging
MYH7	c.3492C>A	p.Asn1164Lys	Missense	1	VUS			-	0	Pathogenic	Benign (94)	Probably damaging	Deleterious	High	Damaging
MYH7	c.3613G>A	p.Glu1205Lys	Missense	1	Published, disease- causing mutation	Pathogenic (1), Uncertain (1)		-	0	Pathogenic	Benign (57)	Probably damaging	Deleterious	High	Damaging
MYH7	c.4259G>A	p.Arg1420Gln	Missense	1	Possible deleterious mutation	Pathogenic (2), Uncertain (1)		-	0	Pathogenic	Benign (43)	Probably damaging	Deleterious	High	Damaging
MYH7	c.4283T>C	p.Leu1428Ser	Missense	1	Possible deleterious mutation	Pathogenic (1), Uncertain (1)		0.00329% (4/121404)	0	Pathogenic	Pathogenic (144)	Probably damaging	Deleterious	Medium	Damaging
MYH7	c.5302G>A	p.Glu1768Lys	Missense	1	VUS	Pathogenic (1), Uncertain (1)		-	0	Pathogenic	Benign (57)	Probably damaging	Deleterious	High	Damaging
MYH7	c.5344A>G	p.Met1782Val	Missense	1	Possible deleterious	Uncertain (2)		-	0	Pathogenic	Benign (22)	Probably	Deleterious	Medium	Tolerated

					mutation							damaging			
MYL2	c.103A>G	p.Ile35Val	Missense	1	Novel variant, likely mutation	Pathogenic (1)		-	0	Pathogenic	Benign (30)	Possibly damaging	Neutral	Neutral	Tolerated
MYL2	c.173G>T	p.Arg58Leu	Missense	1	Likely disease-causing mutation	-		-	0	Pathogenic	Pathogenic (102)	Probably damaging	Neutral	Neutral	Damaging
MYL3	c.130-14G>T	p.IVS1-14G>T	Splicing	1	Unknown**	-		0.15265% (184/120538)	-	-	-	-	-	-	-
PRKAG2	c.247C>T	p.Pro83Ser	Missense	1	VUS	Uncertain (1), Benign (1)		-	68	Benign	Benign (73)	Benign	Neutral	Neutral	Tolerated
TNNI3	c.497C>T	p.Ser166Phe	Missense	1*	Deleterious mutation	-		0.00083% (1/119968)	7	Benign	Pathogenic (155)	Possibly damaging	Deleterious	Medium	Damaging
TNNI3	c.586G>A	p.Asp196Asn	Missense	1	Published, disease-causing mutation	Pathogenic (3), Uncertain (1)	24, 35	0.00083% (1/120728)	3	Benign	Benign (23)	Probably damaging	Deleterious	Medium	Damaging
TNNT2	c.257A>C	p.Asp86Ala	Missense	1	Possibly associated mutation	Pathogenic (2)		0.00085% (1/117696)	1	Benign	Pathogenic (126)	Probably damaging	Deleterious	Low	Damaging
TNNT2	c.236T>A	p.Ile79Asn	Missense	1	Deleterious mutation	Pathogenic (6)	46, 47	-	25	Benign	Pathogenic (149)	Probably damaging	Deleterious	Low	Damaging
TNNT2	c.388C>T	p.Arg130Cys	Missense	1	Probable deleterious mutation	-	48	-	0	Pathogenic	Pathogenic (179)	Probably damaging	Deleterious	Medium	Damaging
TNNT2	c.832C>T	p.Arg278Cys	Missense	2	Deleterious mutation**	Pathogenic (3), Uncertain (4)	49	0.04291% (40/93228)	9	Benign	Pathogenic (179)	Probably damaging	Deleterious	Low	Damaging

\* Denotes a variant in a patient also hosting another variant

\*\* Denotes a variant that would be considered benign/VUS-likely benign based on high frequency of variant in ExAC exome data.

