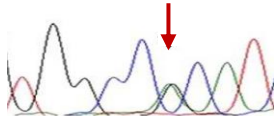


Supplemental file 13.

Figure S5. Sanger sequencing showing gene mutations observed in the present study in patients with VT. Mutation sites are indicated by arrows. Pathogenicity class (ACMG) and #cases - carriers of mutations also shown.

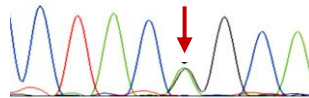
640
T G G C C A C A T



MYBPC3, R160W
c.478C>T, p. Arg160Trp

VUS
iVT #233

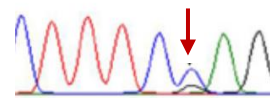
340
C T A C G G C A



MYBPC3, R326Q
c.977G>A, p.Arg326Gln

Benign
CHM VT #226, #391
DCM VT #210, #278

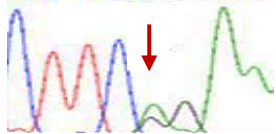
420
C T T T C C A G



MYBPC3, Q998E
c.2992C>G, p.Gln998Glu

VUS
DCM VT #410, #480
iVT #291

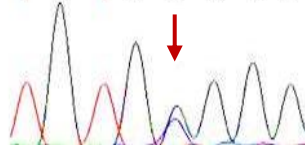
C T T C G G A A



KCNJ2, R218Q
c.653G>A, p.Arg218Gln

Pathogenic
iVT #511

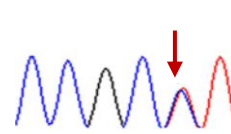
110
T G T G G G G G



GAA, W746C
c.2238G>C, p.Trp746Cys

Pathogenic
CHD VT #533

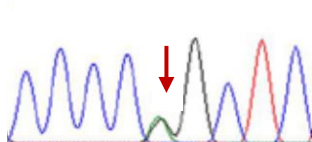
300
C C G C T T



MYH7, F244L
c.730T>C, p.Phe244Leu

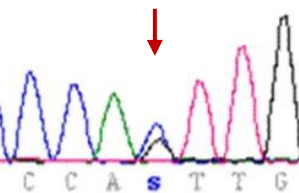
Pathogenic
DCM VT #574

460
C C C C A G C T C



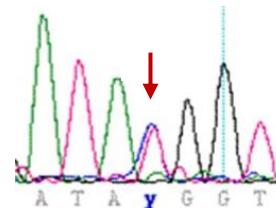
PRKAG2, G100S
c.298G>A, p.Gly100Ser

Benign
CHD VT #344, #436, #412
DCM VT #199
iVT #490



RYR2, V1810L*
c.5428G>C, p.Val1810Leu

Benign
iVT #271



RYR2, T2504M*
c.7511C>T, p.Thr2504Met

Benign
iVT #444

* Akilzhanova A, Guelly C, Nuralinov O, et al. RYR2 sequencing reveals novel missense mutations in a Kazakh idiopathic ventricular tachycardia study cohort. *PLoS One*. 2014;9(6):e101059. Published 2014 Jun 30. doi:10.1371/journal.pone.0101059

Table S7. Sequences of primers used for validation of selected genetic variants.

Gene symbol	Protein/nucleotide change	Exon		Primer (5' to 3')	PCR fragment (bp)
MYBPC3	R160W c.478C>T, p. Arg160Trp	1	Sense:	GCTCCTCTGCTCCCTAC TTCC	708
			Antisense:	ATGGCCATCAGCACAC TTCAC	
MYBPC3	R326Q, c.977G>A, p.Arg326Gln	12	Sense:	GGCGGCACAGAGGGGA TTG	680
			Antisense:	ACCGGCAGGAGCAAAA GGATG	
MYBPC3	Q998E, c.2992C>G, p.Gln998Glu	27	Sense:	TATGTGACCAGTGGGC AGTTC	1,093
			Antisense:	GGGTCTTGTGACTGCA CAAAG	
KCNJ2	R218Q, c.653G>A, p.Arg218Gln	2	Sense:	GGCGAGTGGGCAATCT TCG	470
			Antisense:	CTCAAATCATATAAAG GACTGTC	
GAA	W746C, c.2238G>C,p.Tr p746Cys	16	Sense:	CGTTCAACAAGGATGG CTTC	381
			Antisense:	GTGGGTTCTCCAGCTCA TTG	
MYH7	F244L, c.730T>C, p.Phe244Leu	8	Sense:	CTTGCTGGTCTCCAGTA GTATTGT	536
			Antisense:	GGCTGAGCCTAGCAGA TTCAT	
PRKAG2	G100S, c.298G>A, p.Gly100Ser	3	Sense:	CAGTCCTGTGTGGTCA GAACTTGG	907
			Antisense:	GGACCAGAAGGATTAC GCTTTGAT	
RYR2	V1810L, c.5428G>C, p.Val1810Leu	37	Sense:	TGCGTGCTGGCTACTAT GAC	410
			Antisense:	CCATTACACAGGAATG CAAAGA	
RYR2	T2504M* c.7511C>T, p.Thr2504Met	49	Sense:	GTAACGACGGCCAG TGGCCATTGACACCAA AATTCA	320
			Antisense:	GAAACAGCTATGACCA TGACCCATGGCTTACCT GAAAA	