

SUPPLEMENTAL MATERIAL

Table S1. List of likely pathogenic/pathogenic variants in the recruited individuals.

GnomAD and ClinVar accessed Dec 2019. MAF: minor allele frequency. ACMG: American College of Medical Genetics.

Individual identifier	Gene	Variant	gnomAD[1] MAF	ClinVar [2] classification	Clinical laboratory classification (following ACMG[3] guidelines)
1	<i>TNNI3</i>	c.470C>T p.Ala157Val	Not reported	Pathogenic, two stars	Likely pathogenic
2	<i>MYBPC3</i>	c.2373_2374insG p.Trp792Valfs*41	0.0000174	Pathogenic, two stars	Pathogenic
3	<i>TNNT2</i>	c.305G>A p.Arg102Gln	Not reported	Pathogenic/likely pathogenic, two stars	Likely pathogenic
4	<i>MYBPC3</i>	c.1168delC p.His390Metfs*16	Not reported	Pathogenic, two stars	Pathogenic

5	<i>TPM1</i>	Asp175Asn	0.0000159	Pathogenic, two stars	Likely pathogenic
6	<i>MYBPC3</i>	p.Lys543Argfs*12 c.1628delA	Not reported	Pathogenic, zero stars	Pathogenic
7	<i>MYBPC3</i>	c.1624+4A>T	0.0000133	Pathogenic/lik ely pathogenic, two stars	Pathogenic
8	<i>TNNI3</i>	c.470C>T p.Ala157Val	Not reported	Pathogenic, two stars	Likely pathogenic
9	<i>MYH7</i>	c.1477A>C p.Met493Leu	Not reported	Not reported	Likely pathogenic
10	<i>MYH7</i>	c.1324C>T p.Arg442Cys	0.0000199	Conflicting interpretation, 1 star	Likely pathogenic
11	<i>TNNI3</i>	c.433C>T p.Arg145Trp	0.00000402	Pathogenic, two stars	Likely pathogenic

12	<i>MYBPC3</i>	c.1628delA p.Lys543Argfs*12	Not reported	Pathogenic, zero stars	Pathogenic
13	<i>MYBPC3</i>	c.3163A>T p.Lys1055*	Not reported	Likely pathogenic, one star	Pathogenic
14	<i>MYBPC3</i>	c.1504C>T p.Arg502Trp	0.000401	Conflicting interpretation, one star	Likely pathogenic
15	<i>MYBPC3</i>	c.2096delC p.Pro699Glnfs*55	Not reported	Pathogenic, two stars	Pathogenic
16	<i>MYBPC3</i>	c.1628delA p.Lys543Argfs*12	Not reported	Pathogenic, zero stars	Pathogenic
17	<i>MYBPC3</i>	c.1227-13G>A	0.0000125	Conflicting interpretation, one star	Likely pathogenic
18	<i>MYH7</i>	c.2389G>A p.Ala797Thr	0.0000239	Pathogenic/likely	Likely pathogenic

				pathogenic, two stars	
19	<i>MYBPC3</i>	c.1624+4A>T	0.0000133	Pathogenic/likely pathogenic, two stars	Pathogenic
20	<i>MYBPC3</i>	c.2373_2374insG p.Trp792Valfs*41	0.0000174	Pathogenic, two stars	Pathogenic
21	<i>MYBPC3</i>	c.2373_2374insG p.Trp792Valfs*41	0.0000174	Pathogenic, two stars	Pathogenic
22	<i>MYBPC3</i>	c.3163A>T p.Lys1055*	Not reported	Likely pathogenic, one star	Pathogenic
23	<i>MYBPC3</i>	c.1928-2A>G	Not reported	Pathogenic, two stars	Pathogenic
24	<i>MYBPC3</i>	c.1483C>G p.Arg495Gly	0.00000401	Pathogenic/likely pathogenic, two stars	Likely pathogenic

25	<i>MYH7</i>	c.1711G>A p.Gly571Arg	Not reported	Uncertain significance, one star	Pathogenic
26	<i>TNNT2</i>	c.247G>A p.Glu83Lys	Not reported	Likely pathogenic, one star	Likely pathogenic
27	<i>MYH7</i>	c.2123G>A p.Gly708Asp	Not reported	Not reported	Likely pathogenic
28	<i>TNNI3</i>	c.484C>T p.Arg162Trp	0.0000402	Pathogenic/likely pathogenic, two stars	Likely pathogenic
29	<i>MYBPC3</i>	c.1624+4A>T	0.0000133	Pathogenic/likely pathogenic, two stars	Pathogenic
30	<i>TNNI3</i>	c.484C>T p.Arg162Trp	0.0000402	Pathogenic/likely pathogenic, two stars	Likely pathogenic

31	<i>MYBPC3</i>	c.3293G>A p.Trp1098*	Not reported	Pathogenic, two stars	Pathogenic
32	<i>MYH7</i>	c.2123G>A p.Gly708Asp	Not reported	Not reported	Likely pathogenic
33	<i>MYBPC3</i>	c.2167C>T p.Arg723Cys	Not reported	Not reported	Pathogenic
34	<i>MYBPC3</i>	c.2458C>T p.Arg820Trp	0.00000401	Conflicting interpretation, one star	Likely pathogenic
35	<i>MYBPC3</i>	c.2373_2374insG p.Trp792Valfs*41	0.0000174	Pathogenic, two stars	Pathogenic
36	<i>MYBPC3</i>	c.772G>A p.Glu258Lys	0.0000166	Pathogenic/likely pathogenic, two stars	Likely pathogenic

37	<i>MYBPC3</i>	c.2950C>T p.Gln984*	Not reported	Pathogenic, one star	Pathogenic
38	<i>MYBPC3</i>	c.2373_2374insG p.Trp792Valfs*41	0.0000174	Pathogenic, two stars	Pathogenic
39	<i>MYBPC3</i>	p.Val454Cysfs*12 c.1359delT	Not reported	Pathogenic, one star	Pathogenic
40	<i>MYBPC3</i>	c.1483C>G p.Arg495Gly	0.00000401	Pathogenic/likely pathogenic, two stars	Likely pathogenic
41	<i>MYBPC3</i>	c.1224-19G>A	0.0000256	Likely pathogenic, two stars	Likely pathogenic
42	<i>MYH7</i>	c.1750G>A p.Gly584Ser	Not reported	Pathogenic/likely pathogenic, two stars	Pathogenic

43	<i>MYBPC3</i>	c.1224-19G>A	0.0000256	Likely pathogenic, two stars	Likely pathogenic
44	<i>MYH7</i>	c.1750G>A p.Gly584Ser	Not reported	Pathogenic/likely pathogenic, two stars	Pathogenic
45	<i>MYBPC3</i>	c.(2602+1_2603-1) (3825_?)del NC_000011.9:g.(?_47353422) (47357562_?)del	Not reported	Not reported	Pathogenic
46	<i>CSRP3</i>	c.131T>C p.Leu44Pro	0.000012	Conflicting interpretation, one star	Likely pathogenic
47	<i>MYL2</i>	c.173G>A p.Arg58Gln	0.00000795	Pathogenic/likely pathogenic, two stars	Likely pathogenic
48	<i>MYBPC3</i>	c.1504C>T p.Arg502Trp	0.000401	Conflicting interpretation, one star	Likely pathogenic

49	<i>MYH7</i>	c.2539A>G p.Lys847Glu	Not reported	Likely pathogenic, three stars	Likely pathogenic
50	<i>MYBPC3</i>				

Figure S1. Qualitative and quantitative myocardial hypoperfusion in hypertrophic cardiomyopathy (HCM) mutation carriers in the absence of significant hypertrophy and fibrosis. A. Adenosine stress perfusion maps in the 3 SAX slices, where each pixel encodes myocardial blood flow as per the color scale. B. Corresponding raw stress perfusion imaging. C. Corresponding SAX cine. (perfusion scans are acquired partly in systole). D. Corresponding SAX PSIR LGE imaging. Arrows demonstrate the perfusion defects. LGE = late gadolinium enhancement, PSIR = phase sensitive inversion recovery, SAX = short axis.

















