Supplementary material: detailed relevant genetic findings.

Graziosi M, *et al. Open Heart* 2022; 9:e001914. doi: 10.1136/openhrt-2021-001914

Family	**Patient**	**Diagnostic Pathway**	**CMP phenotype**	**Gene identified**	**Exon**	**Nucleotide Change**	**Protein change**	**Mutation Type**	**Mutation classification According to Varsome https://varsome.com/**	**Mutation Classification According to Clinvar https://www.ncbi.nlm.nih.gov/clinvar/**
I.	1	Screening	N	DSP	10	c.1209C>G	p.Tyr403*	Nonsense	Pathogenic (PVS1,PM2,PP3)	Not reported
I.	2	Screening	N	DSP	10	c.1209C>G	p.Tyr403*	Nonsense	Pathogenic (PVS1,PM2,PP3)	Not reported
I.	3	Screening	N	DSP	10	c.1209C>G	p.Tyr403*	Nonsense	Pathogenic (PVS1,PM2,PP3)	Not reported
I.	4	Screening	HNDC	DSP	10	c.1209C>G	p.Tyr403*	Nonsense	Pathogenic (PVS1,PM2,PP3)	Not reported
I.	5	Screening	N	DSP	10	c.1209C>G	p.Tyr403*	Nonsense	Pathogenic (PVS1,PM2,PP3	Not reported
II.	6	Screening	DCM	SCN5A ANK2	14 38	c.2244G>A c.10054G>C	p.Met748Ile p.Val3352Leu	Missense Missense	Likely Pathogenic (PM1, PM2,PP2,PP3) Likely Benign (PM2,BP1,BP4)	VUS Not reported
II.	7	Screening	HNDC	SCN5A ANK2	14 38	c.2244G>A c.10054G>C	p.Met748Ile p.Val3352Leu	Missense Missense	Likely Pathogenic (PM1, PM2,PP2,PP3) Likely Benign (PM2,BP1,BP4)	VUS Not reported
III.	8	HF	DCM	DSP	11	c.1339C>T	p.Gln447*	Nonsense	Pathogenic (PVS1,PM2,PP3)	Pathogenic

IV.	9	Screening	N	DSP	23b	c.3533T>G	p.Leu1178Arg	Missense	Likely Pathogenic (PM2,PP3)	Not reported
IV.	10	Sudden death	N	DSP TMEM43	23b 12	c.3533T>G c1150C>G	p.Leu1178Arg p. Leu384Val	Missense Missense	Likely Pathogenic(PM2,PP3) Benign (BS1, BS2, BP1, BP4)	Not reported VUS
V.	11	Arrhythmic	HNDC	DSP	20	c.2821C>T	p.Arg941*	Nonsense	Pathogenic (PVS1,PP5,PM2,PP3)	Pathogenic/Likely Pathogenic
V.	12	Screening	N	DSP	20	c.2821C>T	p.Arg941*	Nonsense	Pathogenic (PVS1,PP5,PM2,PP3)	Pathogenic/Likely Pathogenic
VI.	13	Chest pain	N	DSP	24	c.6496C>T	p.Arg2166*	Nonsense	Pathogenic	Pathogenic
VII.	14	Arrhythmic	N	DSP	6	c. 894G>A	p.Trp298*	Nonsense	Pathogenic (PVS1,PM2,PP3)	Pathogenic
VIII.	15	Heart failure	DCM	DSP	23	c.5342delG	p.Arg1781AsnfsTer12	Frameshift	Pathogenic (PVS1,PM2,PP3)	Not reported
VIII.	16	Screening	N	DSP	23	c.5342delG	p.Arg1781AsnfsTer12	Frameshift	Pathogenic (PVS1,PM2,PP3)	Not reported
IX.	17	Arrhythmic	N	PKP2 TMEM43	7 12	c.1643delG c.1112A>G	Gly548fs*562 pTyr371Cys	Frameshift Missense	Pathogenic (PVS1,PP5,PM2) Likely Benign (PP3.BS2,BP1)	Pathogenic VUS
X.	18	Heart Failure	DCM	DSP	24	c.6553C>T	p.Gln2185*	Nonsense	Pathogenic (PVS1,PM2,PP3)	Not reported

XI.	19	Screening	HNDC	DSP FLNC	23 7	c.3361G>T c.1128C>A	p.Glu1121* p. Asn376Lys	Nonsense Missense	Pathogenic (PVS1,PM2,PP3) VUS (PM2,BP1)	Not reported VUS
XI.	20	Arrhythmic	HNDC	DSP FLNC	23 7	c.3361G>T c.1128C>A	p.Glu1121* p. Asn376Lys	Nonsense Missense	Pathogenic (PVS1,PM2,PP3) VUS (PM2,BP1)	Not reported VUS
XII.	21	Sudden death	N	FLNC	48	c.8034delC	p.Cys2679fsTer6	Frameshift	Pathogenic	Not reported
XII.	22	Screening	DCM	FLNC	48	c.8034delC	p.Cys2679fsTer6	Frameshift	Pathogenic	Not reported
XIII.	23	Chest pain	HNDC	DSP	18	c.2611_2614del GATA	p.Asp871AsnfsTer17	Frameshift	Pathogenic (PVS1,PM2,PP3)	Not reported
XIV.	24	Screening	HNDC	DSP	23	c.3465G>A	p.Trp1155*	Nonsense	Pathogenic (PVS1,PM2,PP3)	Pathogenic
XV.	25	Screening	DCM	DES	1	c.537_554del	p.Glu179_Leu184del	Inframe Deletion	Likely Pathogenic (PM1,PM2,PM4.PP3)	Not reported
XVI.	26	Screening	DCM	DSP	24	c.7000C>T	p.Arg2334*	Nonsense	Pathogenic (PVS1,PM2,PP3)	VUS
XVII.	27	Arrhythmic	HNDC	DSG2	13	c.1912G>A	p.Gly638Arg	Missense	VUS (PM2,PP3,BP1)	VUS
XVII.	28	Screening	DCM	DSG2	13	c.1912G>A	p.Gly638Arg	Missense	VUS (PM2,PP3,BP1)	VUS

XVIII.	29	Chest pain	DCM	DSP	4	c.448C>T	p.Arg150*	Nonsense	Pathogenic (PVS1,PM2,PP3)	Not reported
XIX.	30	Chest pain	N	DSP	24	c.6850C>T	p.Arg2284*	Nonsense	Pathogenic (PVS1,PM2,PP3,PP5)	Pathogenic
XX.	31	Arrhythmic	CMP	RYR2	40	c.6289G>A	p.Val2097Met	Missense	VUS (PM1,PM2,PP3)	Not reported
XXI.	32	Arrhythmic	DCM	FLNC	42	c.7000G>T	p.Glu2334Ter		Pathogenic	Not reported

#VarSome: The Human Genomic Variant Search Engine. Christos Kopanos, Vasilis Tsiolkas, Alexandros Kouris, Charles E. Chapple, Monica Albarca Aguilera, Richard Meyer, and Andreas Massouras. *Oxford Bioinformatics*, bty897, 30 October 2018. doi: https://doi.org/10.1093/bioinformatics/bty897