

Supplementary Table 1 – Anatomopathological data from the literature

Manuscript	total number of patients	Source	Cases with biopsy or autopsy	Patient identification	Age (years)	Sex	Variant	Heart weight in autopsy (g)	Vacuolization (H&E)	Glycogen (PAS+)	hypertrophy	Fibrosis (Masson's trichrome)	Inflammation	Myocyte disarray or degeneration	Ultrastructural examination of heart
1 Arad M, et al., J Clin Invest 2002;109:357–362	70	Autopsy (sudden death)	1	-	26	-	N488I	-	pronounced	yes	yes	minimal interstitial fibrosis	no	no	-
2 Murphy RT, et al., JACC 2005;45:922-30	45	RV EMB Autopsy (sudden death)	2 1	II:2 ?	39 27	-	T400N N488I	-	profound extensive	yes yes	yes yes	no coarse interstitial fibrosis	no no	no minimal disarray	increased glycogen non specific degenerative changes in mitochondria. Increased glycogen
3 Bayrak F, et al., Eur J Heart Fail 2006;8:712-5	8	RV EMB	1	IV.1	19	F	E506K	-	pronounced	-	yes	minimal interstitial fibrosis	no	no	-
4 Tan HL, et al., Circ Arrhythm Electrophysiol 2008;1:276-81	10	Autopsy (sudden death)	1	III:8	42	F	R302Q	540	slight	yes	yes	slight interstitial fibrosis	no	extensive myocardial disarray (20% LV mass)	-
5 Kelly BP, et al., Pediatr Cardiol 2009;30:1176-9	1	RV EMB	1	-	1.5	M	E506Q	-	no	no	no	no	no	no	normal amount of glycogen
6 Sternick EB, et al., Heart Rhythm 2011;8:58-64	10	RV EMB	1	I F-3	20	M	R302Q	-	profound	yes	yes	no	no	no	-
7 Liu Y, et al., PLoS ONE 2013;8(5):e64603	1	RV EMB	1	II:2	19	M	K485E	-	profound	yes	yes	interstitial fibrosis	no	prominent focal myocyte disarray	increased glycogen
8 Poyhonen P, et al., J Cardiovasc Mag Res 2015;17:89	6	Heart explant - heart transplant	1	I:1	24	M	H344P	-	pronounced	yes	yes	focal	no	no	increased glycogen
9 Thevenon J, et al., Europace 2016;10.1093/europace/euw067	34	RV EMB	1	II:1 F-1	-	F	R302Q	-	pronounced	yes	yes	no	no	no	-

		RV EMB	2	II:1 F-4	-	F	R302Q	-	no	no	yes	no	no	no	-
		Heart explant - heart transplant	3	III:1 F-9	-	F	H530R	373	pronounced	-	yes	interstitial fibrosis	no	myofibrillar disorganization	-
10	Yogasundaran H, et al., Circ Heart Fail 2016;9:e003367	RV EMB	1	-	31	M	R302Q		pronounced	yes	yes	interstitial fibrosis	no	no	increased glycogen and mitochondrial changes
11	Back Sternick E, et al., Circ Arrhythm Electrophysiol 2016;9:e004455	RV EMB	1	-	46	M	R302Q	-	pronounced	yes	yes	no	no	no	large cytosolic pools of glycogen
		R Atrial EMB	1	-	-	-	-	-	pronounced	yes	yes	no	no	no	large cytosolic pools of glycogen
12	Van der Steld LP, et al., Am J Case Rep 2017;18:766-77	Autopsy (sudden death)	1	-	22	M	K290I	786	severe	yes	biventricular	interstitial fibrosis	no	no	increased glycogen
13	Bhanankha P, et al., BMC Med Genet 2018;19:1-4	RV EMB	1	-	23	F	R302Q	-	pronounced	yes	yes	no	no	no	marked cytoplasmic glycogen accumulation
14	Albernaz Siqueira MH et al., Europace 2020. doi:10.1093/europace/euaa/014/58111872	RV EMB	1	III.1	20	F	H401Q	-	pronounced	yes	yes	no	no	no	large cytosolic pools of glycogen
		RV EMB	2	III.3	18	F	H401Q		pronounced	yes	yes	no	no	no	large cytosolic pools of glycogen
15	Lopes-Sainz A et al., JACC 2020;76(2):186-97	RV EMB	1	-	-	-	R302Q		pronounced	yes	yes	no	no	no	-
16	Hu D, et al., EBioMedicine 2020;54:102723	Autopsy	1	probable	22	M	K485E	1043	pronounced	-	marked	interstitial fibrosis	no	myocyte disarray	large amounts of glycogen
		RV EMB	2	probable	24	M	R302Q		pronounced	-	yes	no	no	partial degeneration	-