

**SUPPLEMENTARY TABLE 2. RARE AETIOLOGIES IN RELATION TO
CARDIOMYOPATHY SUBTYPES ON PROBAND PATIENTS*§**

	Total cohort (N=1929)	HCM (N=1043)	DCM (N=771)	RCM (N=38)	P-value
Malformation syndrome, n(%)	9/1745 (0.5%)	7/907 (0.8%)	2/736 (0.3%)	0/33 (0.0%)	0.535 †
LEOPARD syndrome	2/1745 (0.1%)	2/907 (0.2%)	0/736 (0.0%)	0/33 (0.0%)	
Noonan syndrome	2/1745 (0.1%)	2/907 (0.2%)	0/736 (0.0%)	0/33 (0.0%)	
Other	5/1745 (0.3%)	3/907 (0.3%)	2/736 (0.3%)	0/33 (0.0%)	
Inherited metabolic disorder, n(%)	14/1745 (0.8%)	12/907 (1.3%)	0/736 (0.0%)	2/33 (6.1%)	<0.001 †
Anderson Fabry disease	11/1745 (0.6%)	10/907 (1.1%)	0/736 (0.0%)	1/33 (3.0%)	
Familial amyloidosis	1/1745 (0.1%)	0/907 (0.0%)	0/736 (0.0%)	1/33 (3.0%)	
Other	2/1745 (0.1%)	2/907 (0.2%)	0/736 (0.0%)	0/33 (0.0%)	
Mitochondrial disorder, n(%)	5/1929 (0.3%)	3/1043 (0.3%)	1/771 (0.1%)	1/38 (2.6%)	0.113 †
Kearns Sayre syndrome	1/1929 (0.1%)	0/1043 (0.0%)	0/771 (0.0%)	1/38 (2.6%)	
MELAS syndrome	2/1929 (0.1%)	2/1043 (0.2%)	0/771 (0.0%)	0/38 (0.0%)	
Other	2/1929 (0.1%)	1/1043 (0.1%)	1/771 (0.1%)	0/38 (0.0%)	
Neuromuscular diseases	27/1745 (1.5%)	5/907 (0.6%)	22/736 (3.0%)	0/33 (0.0%)	<0.001 †
Becker MD	8/1745 (0.5%)	0/907 (0.0%)	8/736 (1.1%)	0/33 (0.0%)	
Duchenne MD	3/1745 (0.2%)	0/907 (0.0%)	3/736 (0.4%)	0/33 (0.0%)	
Emery-Dreifuss/Limb-Girdle MD	1/1745 (0.1%)	0/907 (0.0%)	1/736(0.1%)	0/33 (0.0%)	
Friedreich's ataxia	1/1745 (0.1%)	1/907 (0.1%)	0/736 (0.0%)	0/33 (0.0%)	
Myotonic dystrophy	5/1745 (0.3%)	0/907 (0.0%)	5/736 (0.7%)	0/33 (0.0%)	
Other	9/1745 (0.5%)	4/907 (0.4%)	5/736 (0.7%)	0/33 (0.0%)	
Chromosomal abnormalities, n(%)	1/1745 (0.1%)	1/907 (0.1%)	0/736 (0.0%)	0/33 (0.0%)	1.000 †
Down syndrome	1/1745 (0.1%)	1/907 (0.1%)	0/736 (0.0%)	0/33 (0.0%)	
Total, n(%)	54/1929 (2.8%)	27/1043 (2.6%)	24/771 (3.1%)	3/38 (7.9%)	0.100 †

*Related to patients of the Long-Term phase (data could not be merged with those from Pilot phase since categories less detailed and differently described in Pilot study)

† : Fisher exact test S : Significant test

§ No patient with ARVC had a diagnosis of a rare aetiology so ARVC does not appear here

MD : muscular dystrophy