Supplementary Table Table 2

Genetic Variants Co-segregating with the Phenotype Family 039						
Gene	**Variant**			**MAF (1,000G,Eur)**	**CADD-Phred score**	**Expression in cardiac myocytes**
	Ref Seq	**Coding sequence**	**AA change**			
Biologically Plausible						
TTN	NM_003319	c.A9719G	p.K3240R	0.001	18.05	High
	NM_133437	c.18197-1G>A	Splice acceptor	ND	26.9	
Biologically implausible						
ACOX3	NM_001101667	c.A448C	p.M150L	0.010	3.06	Low to Medium
CCDC74A	NM_001258305	c.G688A	p.G230S	ND	0.01	Low
CYP2D6	NM_001025161	c.G941A	p.R314H	ND	34.0	Low
		c.G833T	p.R278L	ND	14.89	
DNAH7	NM_018897	c.G2637T	p.M879I	0.035	3.92	Low
DTX2	NM_001102595	c.C1093T	p.R365C	ND	13.4	Medium
FRG2B	NM_001080998	c.G25A	p.D9N	ND	10.46	Low
FSIP2	NM_173651	c.T17381C	p.M5794T	ND	13.82	Low
GOLGA8A	NM_181077	c.G200C	p.R67P	ND	4.46	Low
GORASP2	NM_001201428	c.C1091T	p.S364F	ND	11.62	Low
HLA-DRB1	NM_002124	c.T397G	p.S133A	ND	0	Low
IL32	NM_001012636	c.A488G	p.D163G	ND	0.81	Low
	ENSG00000008517	c.454dupG	p.Asp152fs	ND	NA	
KRTAP10-7	NM_198689	c.A475G	p.I159V	ND	0.01	Low
LILRA6; LILRB3	NM_001081450	c.T206G	p.L69W	ND	0.01	Low
MUC6	NM_005961	c.C5494T	p.P1832S	ND	11.67	Low
MUC12	NM_001164462	c.G58C	p.V20L	ND	0.13	Low
NBPF14; NBPF26	NM_001351372	c.A592G	p.K198E	ND	0.01	Medium
OR11H12	NM_001013354	c.G596T	p.R199L	ND	0.0	Low
OR2T33	NM_001004695	c.T479C	p.V160A	ND	0.01	Low
		c.T590G	p.M197R	ND	6.23	
		c.T145C	p.W49R	ND	0.05	

PARP4	NM_006437	c.T3116C	p.I1039T	ND	17.08	Low
PDE4DIP	NM_001350520	c.G7358A	p.R2453Q	ND	0.01	Medium to High
	NM_001198832	c.C211A	p.L71I	ND	19.2	
PIK3C2G	NM_004570	c.T3869G	p.V1290G	0.022	0.03	Low
PJVK	NM_001353776	c.G880A	p.G294R	0.025	16.89	Low
POM121	NM_172020	c.G2254A	p.V752I	ND	0.01	Low
		c.T2266C	p.Y756H	ND	0.06	
PRAMEF 1	NM_001294139	c.A423C	p.R141S	ND	5.87	Low
PRB1	NM_001367912	c.G1249A	p.E417K	ND	2.98	Low
RFPL3	NM_001098535	c.C838T	p.R280C	ND	10.28	Low
RGPD5; RGPD8	NM_001164463	c.G4708A	p.G1570R	ND	8.95	Low
RP1L1	NM_178857	c.G3955A	p.A1319T	ND	2.93	Low
TPSD1	NM_012217	c.G274A	p.A92T	ND	0.60	Low
TRIOBP	NM_001039141	c.G1478A	p.S493N	ND	4.19	Medium
TUBA3D	NM_080386	c.C661A	p.R221S	ND	3.46	Low
UBXN11	NM_001077262	c.G1126T	p.G376C	ND	8.13	Low
VCX3A	NM_016379	c.G556A	p.V186M	ND	10.55	Low
ZNF730	NM_001277403	c.C1235T	p.T412I	0.040	6.31	Low
SERHL2	ENSG00000182841	n.588+2dupT	Splice donor-intron	ND	NA	Low

Abbreviations: Gene symbols are per HUGO nomenclature, Ref Seq: Reference sequence; CADD: Combined Annotation Dependent Depletion; AA: amino acid; NA: Not applicable; ND: Not detected. 1000G,Eur: The 1,000 genomes project in the European population.

TTN gene and its variants are listed in bold letter, because of high likelihood pathogenicity.