Family ID	Exon	Nucleotide	Amino Acid	Polyphen-2	Polyphen-2
	Number	Change	Change	Prediction	Score
CHOP-AG ⁶	2	c.175T>C	p.W59R	Probably damaging	1.000
FG-CI ⁵	8	c.784T>C	p.S262P	Probably damaging	1.000
FG-HW	4	c.457T>C	p.F153L	Probably damaging	1.000
FG-HX	2	c.214G>C	p.E72Q	Possibly damaging	0.911
FS-A ⁵	8	c.763A>G	p.K255E	Possibly damaging	0.719
FS-X ⁵	8	c.776C>T	p.T259I	Probably damaging	1.000
FS-XL ⁶	4	c.445- 447delATC	p.I149del	N/A	N/A

Supplementary Table 1. List of *ACTN4* heterozygous missense mutations by family and *in silico* protein function prediction according to Polyphen-2 software. Alterations in nucleotide and amino acid sequence are reported using the following NCBI RefSeq accession numbers: ACTN4 – NM_004924 and NP_004915.

Family ID	Exon	Nucleotide	Amino Acid	Polyphen-2	Polyphen-2
	Number	Change	Change	Prediction	Score
				Probably	
FG-BN ⁷	2	c.808T>A	p.S270T	3	0.999
				damaging	
FG-FQ ⁷	13	c.2683C>T	p.R895C	Probably	1.000
ru-ry	15	C.2003C 1	p.R693C	damaging	1.000
				Probably	
FG-MM	2	c.328T>G	p.N110H	3	1.000
				damaging	
FS-XR ⁷	13	c.2689G>A	n E907V	Probably	0.998
rs-AK	13	C.2009U/A	p.E897K	damaging	0.998
FG 77	10	0.455.4. **	17074	27/4	27/4
FS-Z ⁷	12	c.2455A>T	p.K874-	N/A	N/A

Supplementary Table 2. List of *TRPC6* heterozygous missense mutations by family and *in silico* protein function prediction according to Polyphen-2 software. Alterations in nucleotide and amino acid sequence are reported using the following NCBI RefSeq accession numbers: TRPC6 – NM_004621 and NP_004612.