Table S2. Variants detected via whole-exome sequencing

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| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Gene symbol | Chromosome: position start | HGVSc | HGVSp | Proband  (HCM) | Father  (HCM) | Mother  (normal) | Niece  (HCM) | Related diseases (OMIM number), genetic mode |
| MYBPC3 | chr11:47,357,427 | c.2737+1(IVS26) G>T | exon 26 skip | heterozygous | heterozygous | Wild | heterozygous | CMH4(OMIM:115197), AD |
| ACTN2 | chr1:236,849,999 | c.26(exon1) A>G | p.Gln9Arg | heterozygous | heterozygous | Wild | Wild | CMD1AA(OMIM:612158), AD |
| PSEN2 | chr1:227,076,603 | c.640(exon8) G>T | p.Val214Leu | heterozygous | Wild | heterozygous | heterozygous | CMD1V(OMIM:613697), AD |

HGVSc, Human Genome Variation Society coding sequence name; HGVSp, Human Genome Variation Society protein sequence name; ExAC, Exome Aggregation; CMH4, Familial hypertrophic cardiomyopathy type 4; CMD1AA, Dilated cardiomyopathy type 1AA; CMD1V, Dilated cardiomyopathy type 1V; AD, Autosomal dominant.