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gene mutations and investigate the genotype-phenotype correlation in 10 Chinese
pedigrees with familial hypertrophic cardiomyopathy (HCM). METHODS: There are 91
family members from these 10 pedigrees and 5 members were normal mutated
carriers, 23 members were HCM patients (14 male) aged from 1.5 to 73 years old.
The functional regions of myosin heavy chain gene (MYH7), cardiac myosin-binding
protein C (MYBPC3) and cardiac troponin T gene (TNNT2) were screened with PCR
and direct sequencing technique. Clinical information from all patients was also
evaluated in regard to the genotype. RESULTS: Mutations were found in 5 out of
10 pedigrees. Mutations in MYH7 (Arg663His, Glu924Lys and Ile736Thr) were found
in 3 pedigrees and 3 patients from these pedigrees suffered sudden death at age
20-48 years old during sport. Mutations in MYBPC3 were found in 2 pedigrees, 1
with complex mutation (Arg502Trp and splicing mutation IVS27 + 12C > T) and 1
with novel frame shift mutation (Gly347fs) and the latter pedigree has sudden
death history. No mutation was identified in TNNT2. CONCLUSIONS: Although the
Han Chinese is a relatively homogeneous ethnic group, different HCM gene
mutations were responsible for familiar HCM suggesting the heterogeneity nature
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risk of sudden death in this cohort. Furthermore, identical mutation might result in different phenotypes suggesting that multiple factors might be involved in the pathogenesis of familial HCM.</rdfs:comment>

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<a:SubjectPosition>1:2</a:SubjectPosition>
<a:Object>myosin heavy chain gene ( MYH7 )</a:Object>
<a:ObjectPosition>4:10</a:ObjectPosition>
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)</a:Subject>
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the genotype</c:label>
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<c:label>Mutations in MYH7 ( Arg663His , Glu924Lys and Ile736Thr ) were found in
3 pedigrees and 3 patients from these pedigrees suffered sudden death at age 20-
48 years old during sport</c:label>
<a:hasKeyterm>
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<c:label>MYH7</c:label>
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<b:topic rdf:resource="http://purl.org/commons/record/uniprotkb/P04461"/>
<b:topic rdf:resource="http://purl.org/commons/record/uniprotkb/P11778"/>
<b:topic rdf:resource="http://purl.org/commons/record/uniprotkb/P12883"/>
<b:topic rdf:resource="http://purl.org/commons/record/uniprotkb/P13540"/>
<b:topic rdf:resource="http://purl.org/commons/record/uniprotkb/P49824"/>
<b:topic rdf:resource="http://purl.org/commons/record/uniprotkb/P79293"/>
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ids="7"</a:KeyInfo>
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<a:SubjectPosition>0</a:SubjectPosition>
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<a:Verb>were</a:Verb>
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<a:SubjectPosition>16:17</a:SubjectPosition>
<a:Object>these pedigrees</a:Object>
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<a:SubjectPosition>16:20</a:SubjectPosition>
<a:Object>sudden death</a:Object>
<a:ObjectPosition>22:23</a:ObjectPosition>
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<a:Preposition>at</a:Preposition>
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<a:hasSubjectRelation rdf:resource="svo_16630449s6_5"/>
<a:Subject>suffered sudden death</a:Subject>
<a:SubjectPosition>21:23</a:SubjectPosition>
<a:hasObjectRelation rdf:resource="svo_16630449s6_7"/>
<a:Object>age 20-48 years old during sport</a:Object>
<a:ObjectPosition>25:30</a:ObjectPosition>
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<a:SubjectPosition>28</a:SubjectPosition>
<a:Object>sport</a:Object>
<a:ObjectPosition>30</a:ObjectPosition>
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<a:hasSentence>
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</rdf:Description>
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<c:label>Mutations in MYBPC3 were found in 2 pedigrees , 1 with complex mutation
( Arg502Trp and splicing mutation IVS27 + 12C &gt; T ) and 1 with novel frame
shift mutation ( Gly347fs ) and the latter pedigree has sudden death
history</c:label>
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<b:topic rdf:resource="http://purl.org/commons/record/uniprotkb/Q14896"/>
<b:topic rdf:resource="http://purl.org/commons/record/uniprotkb/Q90688"/>
<a:KeytermPosition>2</a:KeytermPosition>
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<c:label>in 2</c:label>
<a:KeyInfo>ids="37116"</a:KeyInfo>
<b:topic rdf:resource="http://purl.org/obo/owl/CHEBI#CHEBI_37116"/>
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<rdf:type rdf:resource="WzGO"/>
<c:label>death</c:label>
```

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<a:KeyInfo>ids="GO:0016265" onto="biological_process"</a:KeyInfo>
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<a:SubjectPosition>0</a:SubjectPosition>
<a:Object>MYBPC3</a:Object>
<a:ObjectPosition>2</a:ObjectPosition>
</rdf:Description>
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<rdf:Description rdf:about="svo_16630449s7_1">
<a:Verb>were</a:Verb>
<a:VerbPosition>3</a:VerbPosition>
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<a:Subject>Mutations in MYBPC3</a:Subject>
<a:SubjectPosition>0:2</a:SubjectPosition>
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<a:ObjectPosition>0:2</a:ObjectPosition>
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suggesting the heterogeneity nature of the disease-causing genes and HCM MYH7
mutations are associated with a higher risk of sudden death in this
cohort</c:label>
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mutations</a:Object>
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suggesting the heterogeneity nature of the disease-causing genes and HCM MYH7
mutations</a:Object>
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cohort</a:Object>
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be involved in the pathogenesis of familiar HCM</a:Object>
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