OBJECTIVE: The aim of this study was to screen the disease-causing 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 of the disease-causing genes and HCM MYH7 mutations are associated with a higher
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 familiar HCM.

Analysis of MYH7, MYBPC3 and TNNT2 gene mutations in 10 Chinese pedigrees with familial hypertrophic cardiomyopathy and the correlation between genotype and phenotype
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Analysis of MYH7, MYBPC3 and TNNT2 gene mutations in 10 Chinese pedigrees with familial hypertrophic cardiomyopathy and the correlation between genotype and phenotype.

Correlation between genotype and phenotype.
OBJECTIVE: The aim of this study was to screen the disease-causing gene mutations and investigate the genotype-phenotype correlation in 10 Chinese pedigrees with familial hypertrophic cardiomyopathy (HCM).

hypertrophic cardiomyopathy
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The aim of this study
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investigate
the genotype-phenotype correlation in 10 Chinese pedigrees with familial hypertrophic cardiomyopathy (HCM)
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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.
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Clinical information from all patients was also evaluated in regard to the genotype.

Semantically annotated text with named entities and key terms.
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RESULTS: Mutations were found in 5 out of 10 pedigrees.
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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 of the disease-causing genes and HCM MYH7 mutations are associated with a higher risk of sudden death in this cohort.
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CONCLUSIONS: Although the Han Chinese is a relatively homogeneous ethnic group, different HCM gene mutations were responsible for familiar HCM suggesting the heterogeneity nature of the disease-causing genes and HCM MYH7 mutations.
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Furthermore, identical mutation might result in different phenotypes suggesting that multiple factors might be involved in the pathogenesis of familiar HCM.
suggesting different phenotypes might be multiple factors involved in the pathogenesis of familiar HCM that multiple factors might be involved in the pathogenesis of familiar HCM
result in different phenotypes suggesting that multiple factors might be involved in the pathogenesis of familiar HCM.