中文題目：肥厚型心肌病變基因分析

英文題目：Genetic Analysis of Patients with Hypertrophic Cardiomyopathy

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BACKGROUND

Hypertrophic cardiomyopathy (HCM) is the most-common monogenically inherited heart disease, characterized by thickening of the left ventricular wall, contractile dysfunction, and potentially fatal arrhythmias. Hypertrophic cardiomyopathy (HCM) is a major cause of sudden cardiac death, which is caused primarily by pathogenic variants in genes encoding sarcomere proteins. In this study, we screen HCM patients by next-generation sequencing with in-house HCM panel.

METHODS

Forty-eight HCM patients underwent genetic HCM panel screening. Our panel contains MYH7, MYBPC3, TNNT2, TNNI3, MYL2, MYL3, TPM1, ACTC, LAMP2, PLN, PRKAG2, RYR2, GLA, SCN5A. Library preparation was performed by Fludigm Access Array chip followed by MiSeq sequencing. The *data was* analyzed by *CLCbio Genomics Workbench*.

RESULTS

A total of 13 different mutations in TNNT2, SCN5A, PRKAG2, MYBPC3, MYH7 and ACTC1 genes (7 novel and 5 known mutations) were identified. The most frequently mutated genes were MYH7 and MYBPC3. One patient carried double mutations in MYH7 and SCN5A genes. Five mutations (TNNT2:p.Asn73Lys; PRKAG2:p.Gly100Ser; MYH7:p.Ile702Val; ACTC1:p.Leu238Pro; MYBPC3:p.Gln791*) were novel. In silico analysis tools suggested all these five novel mutations as probable pathological.

CONCLUSIONS

These results together with genetic counseling can make early diagnosis and better management in family members at risk of HCM.