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Genotype-Positive Status Is Associated With Poor Prognoses in Patients With Left Ventricular Noncompaction Cardiomyopathy.

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Abstract

Background Left ventricular noncompaction cardiomyopathy (LVNC) is a genetically and phenotypically heterogeneous disease. This study aims to investigate the genetic basis and genotype-phenotype correlations in a cohort of Chinese patients with LVNC . Methods and Results A total of 72 cardiomyopathy-associated genes were comprehensively screened in 83 adults and 17 children with LVNC by targeted sequencing. Pathogenicity of the detected variants was determined according to their prevalence and American College of Medical Genetics and Genomics recommendations. Baseline and follow-up clinical data were collected. The primary end point was a composite of death and heart transplantation. Overall, 42 pathogenic variants were identified in 38 patients (38%), with TTN , MYH 7, MYBPC 3, and DSP being the most commonly involved genes. At baseline, genotype-positive adults had higher rates of atrial fibrillation and family history, and lower left ventricular ejection fraction, compared with genotype-negative adults. During a median follow-up of 4.2 years, more primary end points occurred in genotype-positive adults than in genotype-negative adults (50.0% versus 23.5%; P=0.013). Multivariable analysis demonstrated that genotype-positive status was associated with higher risks of death and heart transplantation, independent of age, sex, and cardiac function at baseline in patients with LVNC (adjusted hazards ratio, 2.49; 95% confidence interval, 1.15-5.37; P=0.020). Conclusions Our study revealed a distinct genetic spectrum in Chinese patients with LVNC , with variants in TTN , MYH 7, MYBPC 3, and DSP being the most common. The presence of pathogenic variants is an independent risk factor for adverse outcomes and may aid in risk stratification in adult patients. Larger studies are needed to confirm these findings.

KEYWORDS: genetics; left ventricular noncompaction; prognosis

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