Familial Hypertrophic Cardiomyopathy-10 Caused by MYL2 de novo Mutation: A Case Report and Literature Review

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Introduction: To summarize the clinical manifestations and gene mutations of familial hypertrophic cardiomyopathy-10 caused by MYL2 gene mutation with a case report and literature review.

Methods: The clinical features of the patient who was admitted to the Cardiology Department in Shenzhen Children's Hospital in June 2019 and then diagnosed with familial hypertrophic cardiomyopathy-10 caused by MYL2 de novo mutation were analyzed. Related literature was searched at OMIM, PubMed, ClinVar, CNKI and Wanfang database (from the establishment of database to July 2019) by using “MYL2” and “hypertrophic cardiomyopathy” as keywords. The characteristics of MYL2 gene mutation and the clinical phenotype of children with familial hypertrophic cardiomyopathy-10 were summarized.

Result: The boy, 2 years and 6 months old, was sent to hospital because of recurrent syncope. Electrocardiogram showed third-degree atrioventricular block. Cardiac hypertrophy with bialtrial hypertrophy, pulmonary hypertension and tricuspid regurgitation were detected by echocardiography. Cardiac catheterization revealed the pressure of vena cava, right atrium, right ventricle and pulmonary artery to be significantly increased. A de novo heterozygous frame shift mutation of MYL2 gene exon 7 (c.484G>A, p.G162R) was identified with whole exome-sequencing analysis. Left ventricular epicardial single chamber permanent cardiac pacemaker was implanted after diagnosed.

Conclusion: This is the first case report of familial hypertrophic cardiomyopathy-10 with third-degree atrioventricular block as initial symptom caused by mutation of MYL2 gene in Chinese children.