POSTER 126

Hypertrophic cardiomyopathy: genetic factors and their role instratifying the risk for associated outcomes

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Resumo

Introduction: Hypertrophic cardiomyopathy (HCM) is the most common hereditary cardiovascular disease. Sarcomeric genes exhibit a central role in HCM pathogenesis, being related to familial HCM cases [1]. First-degree relatives display a 50% higher risk of developing HCM. However, 40% of the HCM cases show a nonfamilial pattern characterized by the absence of family history or mutations in sarcomeric genes [1,2]. Furthermore, HCM patients exhibit an increased risk of developing several potential outcomes, such as non-sustained ventricular tachycardia, arrhythmias, heart failure, and sudden cardiac death [2]. HCM-related outcomes are the principal cause of morbidity in HCM patients and increase their death risk. Thus, stratifying the risk for these outcomes is of utmost importance for HCM management. **Objectives:** Herein, we aimed to explore the relationship between genetic factors and the development of HCM-related outcomes, how they predict their occurrence, and the emerging possibilities of using genetic signatures to stratify the risk for associated outcomes. **Methods:** Relevant articles related to HCM and to the risk factors for HCM-associated outcomes published on PubMed (U.S. National Library of Medicine) were revised and included in this study. **Results:** Genetic testing enables an early diagnosis in HCM, namely in first-degree relatives. However, an HCM genetic cause is only identified in 30% of cases [3]. Black HCM patients exhibit a higher risk for some outcomes, such as sudden cardiac death and heart failure [4]. Furthermore, patients harboring sarcomeric mutations show an increased risk for non-sustained ventricular tachycardia and sudden cardiac death compared to nonfamilial HCM cases [2]. Moreover, HCM is described as a major cause of sudden death in children and young adults (<35 years old). The North American and European models emerged as predictive models for sudden death. However, the North American model reveals limited specificity and the European, low sensitivity [5]. **Conclusions:** Despite the efforts, predicting the development of related outcomes is still a challenge on HCM management. Uncovering new genetic signatures to allow prediction of the occurrence of HCM-related outcomes is of utmost importance. Thus, new approaches to stratify the risk for these outcomes must be developed, allowing a better follow-up for these patients.

Keywords: genetic signature; mutations; hypertrophic cardiomyopathy; genetic variability; risk stratification

References:

[1] Maron BJ. Clinical Course and Management of Hypertrophic Cardiomyopathy. N Engl J Med, 379: 655-668, 2018

[2] Ingles J, Burns C, Bagnall RD, Lam L, Yeates L, Sarina T, Puranik R, Briffa T, Atherton JJ, Driscoll T, Semsarian C. Nonfamilial Hypertrophic Cardiomyopathy: Prevalence, Natural History, and Clinical Implications. Circ Cardiovasc Genet, 10, 2017

[3] Maron BJ, Desai MY, Nishimura RA, Spirito P, Rakowski H, Towbin JA, Rowin EJ, Maron MS, Sherrid MV. Diagnosis and Evaluation of Hypertrophic Cardiomyopathy: JACC State-of-the-Art Review. J Am Coll Cardiol, 79: 372-389, 2022

[4] Ntusi NAB, Sliwa K. Associations of Race and Ethnicity With Presentation and Outcomes of Hypertrophic Cardiomyopathy: JACC Focus Seminar 6/9. J Am Coll Cardiol, 78: 2573-2579, 2021

[5] Veselka J, Anavekar NS, Charron P. Hypertrophic obstructive cardiomyopathy. Lancet, 389: 1253-1267, 2017