THE RELEVANCE OF THE USE OF GENETIC TESTS IN DISEASES OF THE CARDIOVASCULAR SYSTEM

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Genetic testing is important for the management of several inherited cardiovascular diseases, such as cardiomyopathies, arrhythmic disorders, thoracic aortic aneurysms and dissections, and familial hypercholesterolemia. The objective of the present study is to highlight the usefulness of genetic tests in the diagnosis and management of cardiovascular diseases. This is an integrative literature review with a Search for articles published in the last five Years in the Public Medline (PubMed) and Scientific Electronic Library Online (SciELO) databases, using the Health Sciences Descriptors (DeCS): Cardiology, Genetic Testing and Genetic Counseling. In the end, five articles were selected to carry out the work. The results of data collection demonstrated that the majority of cardiovascular disorders that have a genetic component and that begin in early adulthood are inherited in an autosomal dominant manner, even if their expressivity is varied. Thus, genetic tests can be used in several situations, such as aneurysms and familial thoracic aortic dissection, Loyes-Dietz syndrome, Marfan syndrome, Brugada syndrome, long QT syndrome and cardiomyopathies. However, the use of this resource is still limited, and a family history of at least three generations is important, in addition to appropriate phenotyping for the disease for its implementation. The interpretation of altered genetic variants must be careful, with attention to more up-to-date approaches, including the need for a multidisciplinary approach, with a union between a cardiologist and a geneticist. It is also recommended that the geneticist be included in this research even before carrying out the genetic test, promoting the patient's genetic counseling and necessary guidance for the patient's own consent. It is also worth highlighting that the choice of sequencing is very variable, making it possible to analyze a single gene, or genes with greater prevalence, or even large genetic panels. However, patients and healthcare professionals must be informed about the possibility of errors in test results, such as inconclusive reports and the possibility of mutations in genes that were not included in that analysis. However, it is known that most of the variants responsible for generating cardiomyopathies are missense mutations in MYH7 and MYBPC3, which constitute excellent avenues of analysis for genetic testing in patients with suggestive clinical history and family history. Furthermore, gene screening for cardiomyopathy is relevant, as it is a completely manageable disease, with established treatment, which reduces mortality and improves quality of life. It is worth highlighting those genetic tests are important not only for diagnosis, but also to assess the prognosis of the ongoing pathology. Therefore, it is possible to infer those technological advances have allowed expanding the applicability of genetic tests, which are necessary, together with epigenetic and environmental factors, to explain clinical manifestations of a certain disease, such as hypertrophic cardiomyopathy.

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