

# Brazilian Scientific Production about Cardiomyopathies: A Review from 2010 to 2022

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By definition, myocardopathies or cardiomyopathies (CM) are heart muscle diseases that cause structural and functional myocardial abnormalities in the absence of coronary artery disease, hypertension, valvular disease, and congenital heart disease.<sup>1,2</sup>

CM constitute a very heterogeneous group of heart diseases and are one of the leading causes of death either from end-stage heart failure or sudden death due to arrhythmias.<sup>3</sup> According to the phenotype, CM may be classified into hypertrophic cardiomyopathy, dilated cardiomyopathy, restrictive cardiomyopathy, arrhythmogenic right ventricular cardiomyopathy, and left ventricular noncompaction,<sup>1</sup> where there may overlap of phenotypes.<sup>4</sup> Each phenotype is then subclassified into: genetic origin (familial), non-genetic (non-familial), and mixed, with the genetic form accounting for most of the cases.<sup>4</sup> According to the statement of American College of Medical Genetics and Genomics, of the 59 genes mentioned in the document, 30 (51%) had cardiovascular phenotypes, and 16 (27%) included CM phenotypes.<sup>5</sup> Non-genetic CM may be caused by infections, autoimmune diseases, and endogenous or exogenous myocardial toxicity, whereas genetic CM may be caused by a variety of known mutations and others under study.<sup>6</sup>

Another classification divides CM into primary and secondary. The primary ones affect only or predominantly the myocardium, whereas, in secondary CM, cardiac muscle disease is associated with systemic disorders.<sup>6</sup>

The vast complexity surrounding CM can turn diagnosis and management of these patients challenging. However, since the prevalence of heart failure and potentially fatal arrhythmias is high in patient with CM, it is crucial to correctly identify patients and stratify the risk.<sup>3</sup>

With advances in molecular biology and greater ease of access to genetic tests, the number of patients diagnosed with CM has increased in the last years. Hence, there has been an increase in the number of publications on the topic in the literature. An investigation on the MEDLINE database

for articles from 2010 to 2022 using the search parameters “cardiomyopathy” or “myocardiopathy” found a total of 11,432 articles.<sup>7</sup> After adding the filter “country of affiliation Brazil”, we found 111 articles (1% of total publications). However, the terms cardiomyopathy/myocardiopathy are often used to refer to cardiac disorders that by definition are not cardiomyopathies and may correspond to a great portion of published articles. Therefore, after a review of the 111 articles, 26 were excluded because they did not meet the definition of CM, resulting in 85 articles published by Brazilian researchers in the last years (Figure 1). It is possible to observe a growth in the number of publications on the topic, especially when CM of genetic origin is concerned. From 2010 to 2012, no article on CM of genetic origin was published by Brazilian authors, whereas since 2013, with the advances in research on genetic and molecular basis of the disease,<sup>8</sup> there has been an increase in the number of published articles on the topic (Figure 2).

However, the worldwide contribution of Brazilian researchers on the topic is still minor compared to that of other countries. One of the explanations may be the fact that diagnostic methods, such as genetic testing and cardiac magnetic resonance imaging, are not present in most Brazilian centers. It is also necessary to add that many articles by Brazilian authors may have been published in non-indexed journal that thus are not in the MEDLINE database.

Current advances in genetic sequencing and in non-invasive tests such as cardiac magnetic resonance imaging, will allow for the early diagnosis and treatment of patients with CM. Therefore, the number of patients previously diagnosed with idiopathic CM will decrease, with a consequent increase in the number of patients diagnosed with genetically-based CM. Understanding on the topic is relevant not only to specialists in heart failure but also to generalist cardiologists, who will more often treat these patients.

Brazil has a highly multiracial population who, therefore, may carry numerous mutations. Scientific evidence helps to support and conduct studies on the different phenotypes described here. Despite all difficulties, Brazilian publications have been on the rise. Sharing knowledge through experience contributes to clinical improvement, health education, and changes in the global health context.

## Keywords

Cardiomyopathies; Heart Failure; Sudden Death

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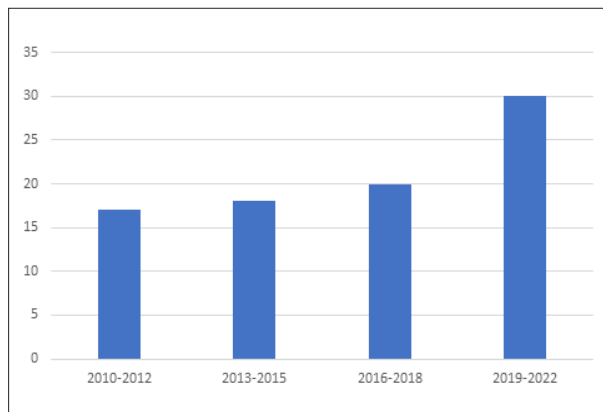
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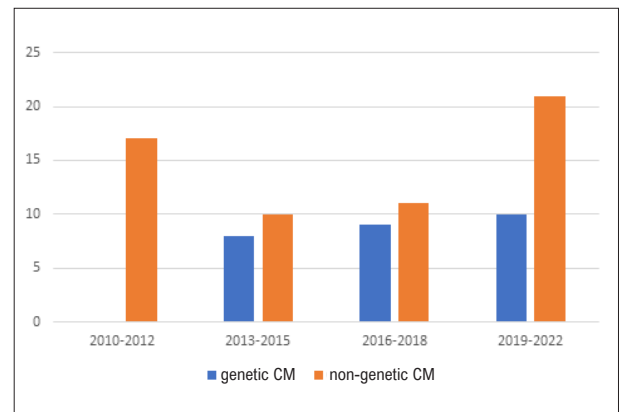
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Conception and design of the research; Acquisition of data; Analysis and interpretation of the data and Writing of the manuscript: Mol EM, Grobe SF; Critical revision of the manuscript for important intellectual content: Moura LAZ, Rohde LEP.



**Figure 1** – Number of articles published by Brazilian researchers from 2010 to 2022.



**Figure 2** – Number of articles published by Brazilian researchers according to the etiological type of cardiomyopathy (genetic CM versus non-genetic CM)

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### Ethics approval and consent to participate

This article does not contain any studies with human participants or animals performed by any of the authors.

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