





# Genetic positive cardiomyopathies: family approach and genetic risk analysis in children

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Cardiomyopathies are diseases of the heart muscle that cause structural and functional changes in the heart. They can affect the heart muscle by weakening it, enlarging it, or altering its structure, which can lead to a reduced ability of the heart to function. There are several main types of cardiomyopathies, and the causes can vary. Cardiomyopathies often do not manifest symptoms in the early stages until serious complications arise, such as heart failure or arrhythmias. Early detection and treatment are crucial for reducing the risk of severe outcomes. We distinguish several types of cardiomyopathies: dilated cardiomyopathy (DCM), hypertrophic cardiomyopathy (HCM), restrictive cardiomyopathy, arrhythmogenic right ventricular cardiomyopathy (ARVC), and Takotsubo cardiomyopathy (stress-induced cardiomyopathy). A common feature of most cardiomyopathies is their genetic cause. Genetically caused cardiomyopathies represent a group of hereditary heart diseases that are transmitted autosomal dominantly or recessively, depending on the type of mutation, from parents to children. These diseases result from mutations in genes that control the structure and function of the heart muscle and can manifest in different forms and vary in disease severity. Genetic factors play a key role in several major types of cardiomyopathies, most commonly in HCM, ARVC, and sometimes in DCM. Genetic testing plays a crucial role in early diagnosis, allowing for the identification of mutation carriers, as well as monitoring and prevention of these diseases in family members predisposed to these diseases.<sup>1</sup> This paper presents the basic facts about genetic cardiomyopathies, along with an overview of the key types of these hereditary heart diseases, with a special focus on their mechanisms of development, clinical manifestations, and diagnostic methods. Additionally, several cases from practice are presented, illustrating the different forms and progression of genetic cardiomyopathies, with the aim of better understanding their impact on patient health and the options for their treatment and management.

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## LITERATURE

1. Lopes LR, Garcia-Hernández S, Lorenzini M, Futema M, Chumakova O, Zateyshchikov D, et al. Alpha-protein kinase 3 (ALPK3) truncating variants are a cause of autosomal dominant hypertrophic cardiomyopathy. *Eur Heart J.* 2021 Aug 21;42(32):3063-3073. <https://doi.org/10.1093/eurheartj/ehab424>