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The era of genes: The importance of genetic research for stratification and therapeutic choice for primary prevention of sudden death in non-ischemic heart failure

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Active military patient, 22 years old, presenting with progressive dyspnea until minimal exertion, associated with paroxysmal nocturnal dyspnea and orthopnea, admitted with heart failure with reduced ejection fraction (EF) (admission echocardiogram EF 09% by Teichozl) hemodynamic profile B and electrocardiogram with QRS < 130ms, without intraventricular conduction disturbance. A diagnostic investigation was carried out after compensating and optimizing the clinical picture. There was a positive family history of dilated cardiomyopathy in the mother at the age of 25. Cardiac magnetic resonance (cMRI) showed late enhancement with a myocardial injury pattern of non-ischemic origin with EF 10%. Six months after admission and optimized therapy, a new MRI showed EF 21%, and ergo spirometry with VO2 max of 35.36ml/kg/min. He conducted genetic research that showed the presence of heterozygosity of the variant described as NM_oo1458.5 ((FLNC):c.6976C>T;p. (Arg2326Ter), classified as pathogenic, in the FLNC gene (Filamin C), (OMIM: 617047), associated with Familial Cardiomyopathy of autosomal dominant inheritance. In young patients with heart failure, in addition to ischemic and infectious etiologies, hereditary etiologies should be considered to stratify the risk of sudden death better. At this point, genetic research provides important information regarding the definitive diagnosis and therapeutic choice. In the case presented, we opted for the implantation of an endocardial cardiac defibrillator for the primary prevention of sudden death, as the patient was a young male with high-risk genetic alterations related to severe heart failure and myocardial fibrosis.