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Titin truncating mutation causing familial dilated cardiomyopathy

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Introduction: Dilated Cardiomyopathy (DCM) is the third most common cause of heart failure among adults. The dilation of heart chambers leads to systolic dysfunction, hence the inability to meet the body's metabolic demands. Familial DCM accounts for 20% of all DCM and is due to Titin (TTN) gene mutations leading to an abnormally truncated Titin protein. Early identification of risk factors and meticulous inquiry regarding family history can lead to earlier identification of the mutation and diagnosis with prompt treatment.

Case Report: A 56 year old male, with an extensive family history of cardiovascular disease, presented with shortness of breath and palpitations. Exercise stress test showed grossly abnormal findings which warranted a transthoracic echocardiogram (TTE) and coronary angiography. TTE depicted mild cardiomyopathy with an ejection fraction (EF) of 50-55% and coronary angiography showed no coronary artery disease. He was subsequently started on Lisinopril and Carvedilol. Approximately 7 years later he presented with progressive shortness of breath and wheezing at an annual checkup. A repeat TTE, showed progression of his cardiomyopathy with EF of 35-40% and diffuse hypokinesis. An implantable cardioverter defibrillator was placed for primary prevention and genetic testing/counselling was discussed. He tested positive as a heterozygous carrier for a mutation that truncates the Titin protein at amino acid 18,386 leading to a frameshift mutation and a shortened protein. The patient was advised that each of his children would have a 50% chance of inheriting the at risk allele. After thorough discussion he decided to have his two children tested, the results of which are still pending.

Discussion: Familial DCM shows an autosomal dominant inheritance, and is genetically heterogeneous. It is estimated that the frequency of TTN truncations affecting the general population is 0.36% and prevalence of frameshift mutations in the A band region is 0.057%. Titin, one of the largest proteins in the body is a crucial component of the cardiac myocyte. Abnormal Titin protein leads to defective contraction of heart muscle leading to heart failure. This case highlights a rare cause of heart failure with reduced ejection fraction, which should be included in the differential diagnosis of all patient's with a non-ischemic cardiomyopathy.

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