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Letter To The Editor

Noncompaction Requires Family Studies, Work-Up for Neuromuscular Disorders, and Close Monitoring

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In a recent article Erdogan *et al.*, reported about a 55yo male with sudden onset anginal chest pain and mild systolic dysfunction in whom left ventricular hypertrabeculation / noncompaction (LVHT) was detected on echocardiography and anginal chest pain attributed to myocardial bridging of the posterior descending artery on multidetector computed tomography (Erdogan, E. *et al.*, 2013). We have the following comments and concerns.

Though LVHT is regarded as a congenital disorder in the majority of the cases, it may develop during postnatal life in rare cases as well (acquired LVHT) (Finsterer, J. *et al.*, 2008). Were any previous echocardiographic examinations available and reviewed, particularly intrauterine ultrasound to confirm the congenital nature of LVHT in this patient?

Diagnosing LVHT has significant therapeutic implications not only in terms of heart failure but also in terms of ventricular arrhythmias and cardio-embolic risk (Caliskan, K. *et al.*, 2012). Since LVHT patients carry an increased risk of ventricular arrhythmias with sudden cardiac death and of cardio-embolic events, it is essential to closely monitor these patients for arrhythmias and for the development of intertrabecular thrombus formation. In patients with a cardio-embolic event and LVHT, oral anticoagulation is indicated. Was the individual history positive for stroke /embolism, did the patient ever undergo cerebral MRI to look for previous embolic ischemias?

Since LVHT frequently occurs also in other family members, it is worthwhile to carry out family studies in case LVHT is diagnosed. Interestingly, relatives of LVHT patients may also present with cardiomyopathy other than LVHT. Were echocardiographies carried out in first-degree relatives of the described patient?

Since the patient presented with two presumably congenital abnormalities and since LVHT is frequently associated with chromosomal abnormalities or mutations in genes encoding proteins of the contractile apparatus or the sarcolemma (Finsterer, J. 2009), it would be interesting to know if investigations for chromosomal aberrations were carried out. Was consanguinity excluded in the family of the patient?

Since LVHT is even more frequently associated with neuromuscular disorders there is a need to refer these patients to the myologist for neuromuscular evaluation. Were there any clinical indications, such as muscle weakness or wasting, muscle cramping, easy tiredness, joint contractures or gait disturbance, for neuromuscular disease in the presented patient? It is also essential to ask LVHT patients for sensory disturbances since it has been described even in association with hereditary neuropathy (Corrado, G. *et al.*, 2006).

So far, LVHT has been reported in association with mutations in the TAZ, DTNA, ZASP, lamin A/C, MYH7, MYH7B, ACTC1,TNNT2, TNNI3, MYBPC3, TPM1, dystrophin, DMPK, ZNF9, LAMP2, GAA, mtDNA genes, AMPD1, GBE1, RYR1, COL7A1, PMP22, MMACHC, β-globin, and DNAJC19 genes.

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Was he tested for mutations in any of these genes? Was the family history positive for hereditary disease?

Overall, this interesting case needs further individual and familial investigations to address these issues raised above and close monitoring to prevent fatal complications of LVHT.

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