LETTER TO THE EDITOR

Noncompaction in Fabry’s disease

Não compactação na doença de Fabry

To the Editor,

We read with interest the article by Martins et al. about the first patient reported with X-linked Fabry’s disease associated with left ventricular hypertrabeculation/noncompaction (LVHT).\(^1\) We have the following comments and concerns.

There is a long-term debate about the pathogenesis of LVHT.\(^2\) Most studies indicate that LVHT derives from an early defect of embryonic cardiac development.\(^3\) However, there are individual cases in which LVHT was not present on previous echocardiographic investigations and was thus assessed as ‘acquired LVHT’, unlike the more common congenital variant.\(^4\) Did the patient presented undergo previous echocardiographic or cardiac MRI investigation? Was LVHT present on these previous studies?

LVHT is associated with a large number of mutated genes but a causal relation between LVHT and associated mutations has not been proven. To assess the relationship between the mutation and LVHT it could be helpful to investigate other family members for LVHT and Fabry’s disease. Since LVHT occurs in families other family members should be always investigated for LVHT. If other family members present with LVHT and Fabry’s disease as well, a causal relation becomes quite likely. A further argument for a causal relation is that Fabry’s has been reported in association with hypertrophic cardiomyopathy,\(^5\) both apical\(^6\) and obstructive\(^7\) types. How certain are the authors of the causal relation between the p.R220X mutation in the GLA gene and LVHT in the patient presented?

LVHT is also frequently associated with neuromuscular disorders\(^8\) and Fabry’s disease manifests in the muscle as well due to glycosphingolipid accumulation in myocytes and muscular arteries.\(^9\) Was there any evidence of muscle disease in the described patient? Were there clinical indications of muscle disease such as muscle weakness, wasting, cramp-}

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Conflicts of interest

The authors have no conflicts of interest to declare.

References


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