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Letter to Editor

Desminopathy due to the DES variant c.735+1G>T manifesting as myopathy and non-compaction

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With interest we read the article by Fan et al. about a Chinese family with desminopathy due to the DES variant c.735+1G>T manifesting with restrictive cardiomyopathy in the index patient (III/14) and with left ventricular hypertrabeculation (LVHT)/no compaction in family member IV/8 [1].

Key words: cardiac involvement; desmin; cardiomyopathy; arrhythmias; non-compaction.

We have the following comments and concerns.

It remains unclear how many of the mutation carriers were neurologically and cardiologic ally investigated and by which means. Since LVHT occurs familiarly [2], we should be informed how many mutation carriers underwent ECG, echocardiography, or cardiac MRI (cMRI), and in how many LVHT was truly detected. Were neurologists and cardiologists aware of the genetic diagnosis?

LVHT might be over diagnosed or overlooked by imaging techniques and the interobserver agreement of the echocardiographic diagnosis is poor [3]. Thus, we should know the criteria according to which LVHT was diagnosed by echocardiography and cMRI. Was LVHT in patient IV/8 also diagnosed by echocardiography? Was cMRI carried out in the index patient?

Since the index patient manifested already at age 8y in the myocardium with left bundle branch block (LBBB), since LVHT may disappear in single cases over time [4], and since LVHT may be diagnosed already on fetal echocardiography [5], we should know if LVHT was present in the index patient on fetal echocardiography, at age 8y, or later, and if it disappeared thereafter.

Figure 3 shows the cMRI of individual VI/8 [1]. However, no individual with this notification can be identified in the pedigree. Is it conceivable that the authors mean individual IV/8? Furthermore, it is unclear which individual's echocardiography and cardiac CT is shown in figure 3A respectively 3B. Cardiac CT shown in figure 3B suggests LVHT as well. If echocardiography and cardiac CT are from

the index patient, we should know why echocardiography is not indicative of LVHT.

Since the index patient had a history of syncope, we should know if it was attributable to ischemic stroke, cerebral breeding, seizure, carotid artery stenosis, or a cardiac cause. We should also know if the patient fainted only once or repeatedly. Since LVHT may be complicated by arrhythmias and cardioembolism, we should know if there were any indications for atrial fibrillation or intraventricular thrombus formation.

ECG in figure 1A suggests PQ- prolongation [1]. Did the index patient also suffer from first degree atrioventricular block?

The type of cardiomyopathy in patient IV/8 is not specified. Since desminopathy has been reported in association with restrictive cardiomyopathy, we should know if echocardiography or cMRI was indicative of restrictive cardiomyopathy, characterised by biatrial enlargement, a restrictive filling pattern, and preserved systolic function.

The pharmacotherapy of both patients is not described [1]. Did the index patient receive oral anticoagulation when he developed atrial flutter?

Overall, the study could be more meaningful if the number of family members undergoing cardiological and neurological investigations would have been provided, if the results of these investigations would have been described in detail, if the therapeutic management would have been reported, and if some inconsistencies would have been purged.

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