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Letter to the Editor

Is Turner's syndrome associated with cardiomyopathy and myopathy?

Dear Sir,

We read with interest the article by van Heerde et al.[1] on a patient with Turner's syndrome with severe mitral valvar stenosis and consecutive pulmonary hypertension secondary to a parachute-like mitral valve, the left superior caval vein draining into the coronary sinus, and left ventricular hypertrabeculation/non-compaction. The study raises several concerns.

The authors suspect that non-compaction developed during embryogenesis. This assumption would imply that the findings had already been present at the age of 9 years when the patient first underwent open cardiac surgery. The authors give no information that non-compaction was observed at this time, neither intra-operatively, nor during preoperative or postoperative echocardiography, suggesting that the subsequent findings were not congenital but rather acquired. It is possible, of course, that non-compaction was already present at the age of 9 years, but that cardiologists and cardiac surgeons were not sensitized to the existence of the entity at that time, and thus did not look for it with sufficient care.

Since non-compaction has been reported to be associated with neuromuscular disorders in four-fifths of the patients in whom it is found,[2] it is surprising that their patient was not referred to the neurologist to look for a neuromuscular disorder. Since Turner's syndrome has been associated with X-linked muscular dystrophies,[3] it cannot be excluded that the patient suffered from a neuromuscular disorder in addition to the chromosomal aberration. Additionally, it cannot be excluded that there was direct skeletal muscle impairment in Turner's syndrome. Their patient, nonetheless, may also fall into the group of patients in whom non-compaction is not associated with a neuromuscular disorder. Neuromuscular disorders which are frequently associated with non-compaction are Becker's muscular dystrophy, mitochondriopathy, myoadenylate-deaminase deficiency, myotonic dystrophy type 1, Pompe's disease, dystrobrevinopathy, myopathy due to a cypher gene mutation, and Barth syndrome.[4]

It is not clear why the surgeons did not explore the purported non-compaction during replacement of the valve after it has been detected by echocardiography. Though non-compaction is most frequently detected on echocardiography or cardiac resonance imaging, the best method to confirm the abnormality is intra-operative inspection or autopsy. It is surprising that non-compaction was not detected intra-operatively on two occasions. Could it be possible that the echocardiographic findings were false positive ones, since non-compaction was also not reported in the postoperative echocardiography performed 1 year after valvar replacement?

There are no indications that non-compaction is caused by disturbed haemodynamics. It is even unclear if the abnormal finding is a congenital disorder, since it has also been shown to develop during the disease course.[5] Pathogenic theories other than non-compaction of the embryogenic myocardium are "dissection" of the myocardium, frustrated attempts at myocardial hypertrophy, myocardial tearing due to dilation or a metabolic defect, or compensatory hypervascularization.[6]

In conclusion, we emphasize that patients with suspected non-compaction should not only undergo cardiological but also neurological investigations. In the majority of the cases, patients with positive findings also suffer from a neuromuscular disorder.

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Reply

Dear Sir,

Re: Is Turner's syndrome associated with cardiomyopathy and myopathy?

We appreciate that Drs Finsterer and Stöllberger have outlined their experience with patients suffering from myocardial non-compaction associated with various forms of neuromuscular disorders. Dr Finsterer may be right that non-compaction was already present in our patient at the age of 9 years, and that our cardiologists were probably not sensitized to the existence of the entity at that time. All of us are now experiencing that, with better quality imaging provided by a new generation of echocardiographic equipment, more details are disclosed. We believe that the diagnosis should be made primarily by echocardiography. We would not expect that the cardiac surgeon would be able to evaluate the appearance of the left ventricular apex during repair of a malformed mitral valve with fused tendinous cords. There exists an overlap between various forms of abnormal myocardial structure, evidenced also by a non-uniform terminology – non-compaction, left ventricular hypertrabeculation, spongy myocardium. In fact, prominent left ventricular trabeculations can be found in many hypertrophic hearts, and strict echocardiographic diagnostic criterions are therefore warranted for the distinction of non-compaction.[1]

In a recent study,[2] in four-fifths of the patients with non-compaction, an association with neuromuscular disorder was found. Our patient demonstrated no clinical features of a neuromuscular disease up to 17 years of age and had normal values of creatine kinase, this together making the diagnosis of any form of a dystrophinopathy less probable. We would hesitate at this moment to refer all patients with myocardial non-compaction, and absence of clinical features of a muscular disease, for a complete and routine workup to exclude neuromuscular disorders. Almost one-fifth of the patients in the above-mentioned study[2] refused the neurological investigation, being reluctant to undertake the additional studies when they appeared only to have a cardiac problem. We should have a low threshold, nonetheless, for investigation of any possible neuromuscular disorder, starting with a specifically oriented history, and we should be prepared to refer the patient for specialist opinion when indicated. We thank Drs Finsterer and Stöllberger for drawing our attention to this interesting issue, focused on the potential neuromuscular involvement in the setting of myocardial disease.

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