Consider the genetic and myopathic background, familial occurrence, and alternative definitions of left ventricular hypertrabeculation/noncompaction

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To the Editor

We read with interest the article by Ali et al. regarding 19 patients with left ventricular hypertrabeculation/noncompaction (LVHT) who additionally presented with mitral regurgitation (MR) with leaflet retraction [1]. We have the following comments and concerns.

The authors attribute MR in LVHT to elongation or rupture of chordae tendinae due to involvement of the papillary muscles [1]. However, some definitions of LVHT explicitly define LVHT as an abnormality distal to the papillary muscles, thus not involving these structures [2].

The authors mention that in patients with congenital LVHT and MR the left ventricle is thin-walled rather than hypertrabeculated [1]. This is contradictory since LVHT is characterized by hypertrabeculation.

It would be interesting to know which neurological complications occurred in the one patient who underwent closure of a septal defect and developed ventricular arrhythmia post-operatively [1]. Did the patient experience stroke/embolism, epilepsy, or syncope? Did neurological complications completely resolve?

LVHT is frequently associated with mutations in various genes expressed in the skeletal muscle and the myocardium or with chromosomal defects. Which of these well-known genetic defects were present in the investigated cohort?

LVHT may not only be diagnosed echocardiographically according to Jenni's criteria but also according to Stöllberger's or Chin's criteria [3,4]. Did the patients included in this study also fulfill these alternative diagnostic criteria?

LVHT has been shown to occur among family members in up to 27% of the cases [5]. In how many of the study patients were other family members investigated for LVHT and in how many was LVHT detected?

The authors measured the ratio of the non-compacted/compacted layer distal to the papillary

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muscles in the short axis view [1]. How could they be sure that the measurement was distal since the origin of papillary muscles would have been difficult to identify in this projection?

We do not agree with the initial sentence of the discussion that ''improvement of heart failure and ejection fraction (EF) is one of the distinctive clinical features of LVHT'' [1]. Systolic dysfunction is a well-known complication of LVHT, which usually improves only upon appropriate treatment but not spontaneously.

Patients included in this study were recruited from two centers [1]. What was the inter-observer agreement concerning the diagnosis of LVHT? Were all patients diagnosed with LVHT in Khartoum also accepted as LVHT in Riyadh and vice versa?

What was the rationale behind the exclusion of LVHT patients with an EF <45%? Some, but not all, patients with systolic dysfunction develop MR.

The authors state that a distinct feature of LVHT with MR is normal systolic function [1]. Contrary to these statements, the authors mention that 4/19 patients had decreased systolic dysfunction [1]. Did the four patients with systolic dysfunction receive heart failure therapy? Why did two patients not respond to treatment?

Worsening of MR and improvement of EF do not necessarily imply that MR is independent of systolic function. The causal relation between the two parameters could be reciprocal.

Overall, this interesting study could profit from a more thorough analysis of the data and from discussion of the many unsolved issues associated with LVHT in general and in particular with the patients investigated in this study. The term ''hypertrabeculation'' has the advantage that it is descriptive whereas ''noncompaction'' implies a causal mechanism which has never been proven.

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