

Letter to Editor

## Left ventricular hypertrabeculation / noncompaction is not unique in trisomy 22

Josef Finsterer, MD, PhD

Krankenanstalt Rudolfstiftung, Vienna, Austria

Corresponding author: Josef Finsterer, MD, PhD, Krankenanstalt Rudolfstiftung, Postfach 20, 1180 Vienna, Austria, Tel. +43-1-71165-72085; Fax. +43-1-4781711; E-mail: ffigs1@yahoo.de

Received: November 20, 2018; Accepted: December 24, 2018; Published: December 28, 2018

**Key words:** chromosomal, non-compaction, hypertrabeculation, genetics, cardiac, neuromuscular

### Letter to the Editor

In a recent article, Abdelgadir et al. reported about two pediatric patients with non-mosaic trisomy 22 of whom the 5 year-old female presented with left ventricular hypertrabeculation, also known as noncompaction (LVHT).[1] We have the following comments and concerns.

Generally, LVHT is regarded as a congenital abnormality of the left ventricular myocardium characterised by a two-layered structure of the myocardium, consisting of a thin compacted outer (pericardial) and a non-compacted inner (endocardial) layer.[2] The non-compacted layer is characterised by a complex interwoven meshwork of trabeculations, which are separated by deep intertrabecular recesses reaching the compacted layer. LVHT is most frequently located in the apex, and the lateral wall but usually spares the septum.[2] LVHT may be associated with other cardiac abnormalities (non-isolated) or may occur as the sole morphological abnormality (isolated LVHT).[3] Isolated and non-isolated LVHT are frequently associated with ventricular arrhythmias or systolic dysfunction.[3] There are also indications that the frequency of stroke or embolism is increased in LVHT due to thrombus formation within the intertrabecular spaces.[2,3] In the vast majority of the cases LVHT is assumed to be present already at birth but no systematic studies on the prevalence of LVHT in early infancy have been carried out so far.

In the abstract Abdelgadir et al. claim that their report is the first one about a patient with trisomy 22 and LVHT. However, LVHT has been previously described in trisomy 22.[4] In an unborn girl with trisomy 22 Wang et al. described in 2007 LVHT on fetal echocardiography.[4] Contrary to the report by Wang et al., LVHT in Abdelgadir's case was absent on fetal echocardiography.[1] Fetal echocardiography showed only moderate tricuspid regur-

gurgitation and a small pericardial effusion.[1] No other echocardiographies were carried out during gestation. Postnatal transthoracic echocardiography surprisingly revealed LVHT associated with a large atrial septal defect, persistent ductus arteriosus, and moderate tricuspid regurgitation. Comparison between fetal and neonatal echocardiographies suggests that LVHT in the presented patient was acquired. Acquired LVHT has been only rarely described and is associated with neuromuscular disorders (NMDs).[5] To exclude that LVHT was missed on fetal intrauterine echocardiography or hidden during gestation and unmasked during delivery or shortly afterwards, the intrauterine examination should be reviewed. If LVHT is truly absent on this initial examination, the authors should provide an explanation why LVHT developed between the intrauterine echocardiography and the neonatal examination. Isolated LVHT with normal systolic function was still present at age 1y.

In addition to chromosomal abnormalities LVHT is frequently associated with NMDs.[5] Was there any indication for a second trouble such as a NMD in the presented patient? Was the family history positive for NMD? Which were the CK levels and was an electromyogram recorded?

Overall, it should be emphasised that the presented patient had acquired LVHT, that it is not the first report of a patient with trisomy 22 and LVHT, that investigations for NMD should be initiated, and that the patient requires close follow-up to prevent systolic dysfunction or severe arrhythmias.

### References

1. Abdelgadir D, Nowaczyk MJ, Li C. Trisomy 22 mosaicism and normal developmental outcome: Report of two patients and review of the literature. *Am J Med Genet A*. 2013.
2. Engberding R, Yelbuz TM, Breithardt G. Isolated noncompaction of the

- left ventricular myocardium - a review of the literature two decades after the initial case description. *Clin Res Cardiol.* 2007;96:481-8.
3. Finsterer J. Cardiogenetics, neurogenetics, and pathogenetics of left ventricular hypertrabeculation/noncompaction. *Pediatr Cardiol.* 2009; 30:659-81.
  4. Wang JC, Dang L, Mondal TK, Khan A. Prenatally diagnosed mosaic trisomy 22 in a fetus with left ventricular non-compaction cardiomyopathy. *Am J Med Genet A.* 2007;143A:2744-6.
  5. Finsterer J, Stöllberger C, Schubert B. Acquired left ventricular noncompaction as a cardiac manifestation of neuromuscular disorders. *Scand Cardiovasc J.* 2008;42:25-30.

To cite this article: Finsterer J. Left ventricular hypertrabeculation / noncompaction is not unique in trisomy 22. *British Journal of Heart Diseases.* 2018; 1:1.

© Finsterer J. 2018.