

Tetralogy of Fallot, Down's Syndrome, Left Ventricular Noncompaction, and Multiples Thrombi

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ABSTRACT

We report a unique case of left ventricular noncompaction (LVNC), tetralogy of Fallot, and multiples biventricular thrombi in a Down's syndrome. Of interest, speckle tracking analysis detected an abnormal LV myocardial longitudinal deformation, despite the normal ejection fraction and absence of NC at the baseline evaluation.

Key Words: Left ventricular noncompaction, speckle tracking analysis, tetralogy of Fallot, thrombi

INTRODUCTION

Left ventricular non compaction is a rare cardiomyopathy, it has been recently reported in association with other congenital heart diseases. We present an unusual association of left ventricular noncompaction with congenital heart disease and genetic syndrome.

CASE REPORT

We report a case of a 6-year-old Down's syndrome girl diagnosed at birth as a case of tetralogy of Fallot (TOF) who underwent complete repair at the age of 6 months by transannular patch followed at our outpatient clinic.

The evaluations performed from 2009 to 2013 demonstrated a progressive appearance of left ventricular

noncompaction (LVNC) areas in the lateral, septal, and apical walls and a progressive reduction of biventricular systolic function [Figure 1]. The ratio, measured in 2013, between the noncompacted and compacted endomyocardial layers was >2.0 , according with the LVNC echocardiographic diagnostic criteria.^[1]

Of interest, speckle tracking analysis of the echo study, done in 2009, detected an abnormal LV myocardial, longitudinal strain (average = 8% in the apical four chamber view) [Figure 2], despite a normal LV ejection fraction (60%) and the absence of LVNC at the baseline evaluation. During the follow-up, there was a drop in LV ejection fraction (50%) with a persistent stable reduction in LV myocardial longitudinal strain.

At the last visit in 2014, she was asymptomatic, but a two-dimensional Doppler color-flow echocardiography showed severe dilatation of both ventricles with a diffuse

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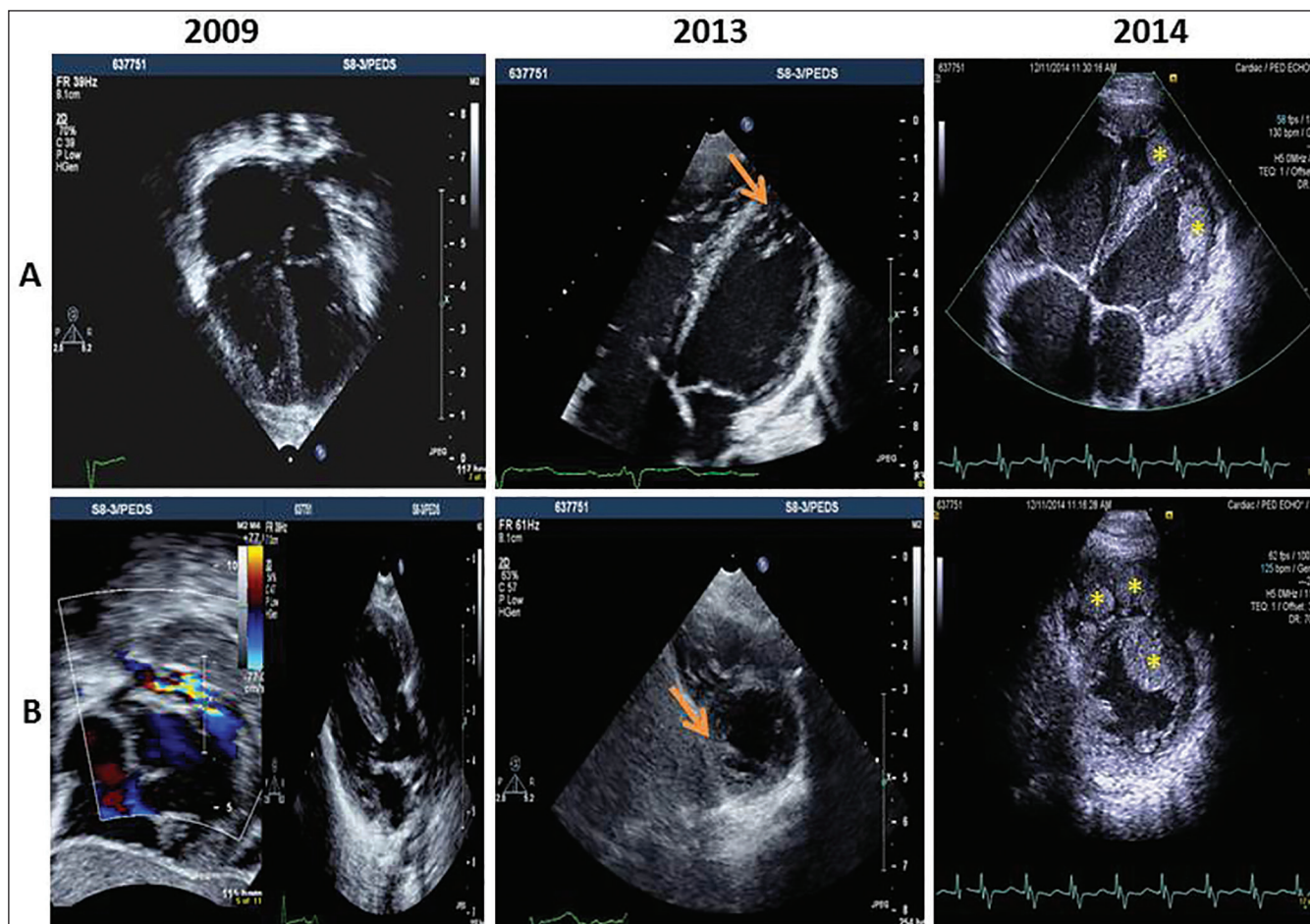


Figure 1: 2009 Panel A: Apical four-chamber view at the baseline evaluation showing no hypertrabeculated left ventricle (LV). 2009 Panel B: Subcostal view and parasternal long axis view showing the typical tetralogy of Fallot features, anterocephalic deviation of the infundibular septum, right ventricular outflow tract obstruction, large ventricular septum defect, overriding aorta. 2013 Panel A: Apical four chamber view showing the presence of hypertrabeculated left ventricular apex (arrow); 2013 Panel B: Parasternal short axis view showing hypertrabeculated LV (arrow). 2014 Panel A: Apical four chamber view showing a clot in the dilated right ventricle (star) and a large clot in the hypertrabeculated LV (star); 2014 Panel B: Parasternal short axis showing multiple clots (stars) in the right ventricle and the hypertrabeculated LV (star)

biventricular hypokinesia, (LV ejection fraction <30%, tricuspid annular plane systolic excursion 8 mm), and free pulmonary regurgitation. Multiple apical biventricular thrombi were detected [Figure 1].

A regimen of heparin (imbricated with warfarin), carvedilol, spironolactone, captopril, and furosemide was started.

DISCUSSION

LVNC is a rare cardiomyopathy due to the arrest of myocardial compaction during embryogenesis that may manifest from infancy to young adulthood.^[1] Standard echocardiography has a well-established role in the diagnosis demonstrating the presence of LV trabeculations apically to the papillary muscles.^[1,2] Previous studies have shown that clots can be formed in LVNC.^[2]

LVNC, originally described as an isolated disease, it has been recently reported in association with other congenital heart diseases (about 12% of LVNC cases).^[3] Particularly, the association with TOF ranges from 4% to 8% in the different series.^[3]

LVNC has been also associated with mutations of several genes or with neuromuscular disorders.^[4] No cases have been reported of LVNC and Down's syndrome.

To the best of our knowledge, this is the first described case of LVNC, TOF, multiples biventricular thrombi, and Down's syndrome.

This case also confirmed the ability of advanced echo modalities such as strain analysis to detect early functional abnormalities in the presence of normal LV ejection fraction and before the appearance of a clear LVNC.^[5]

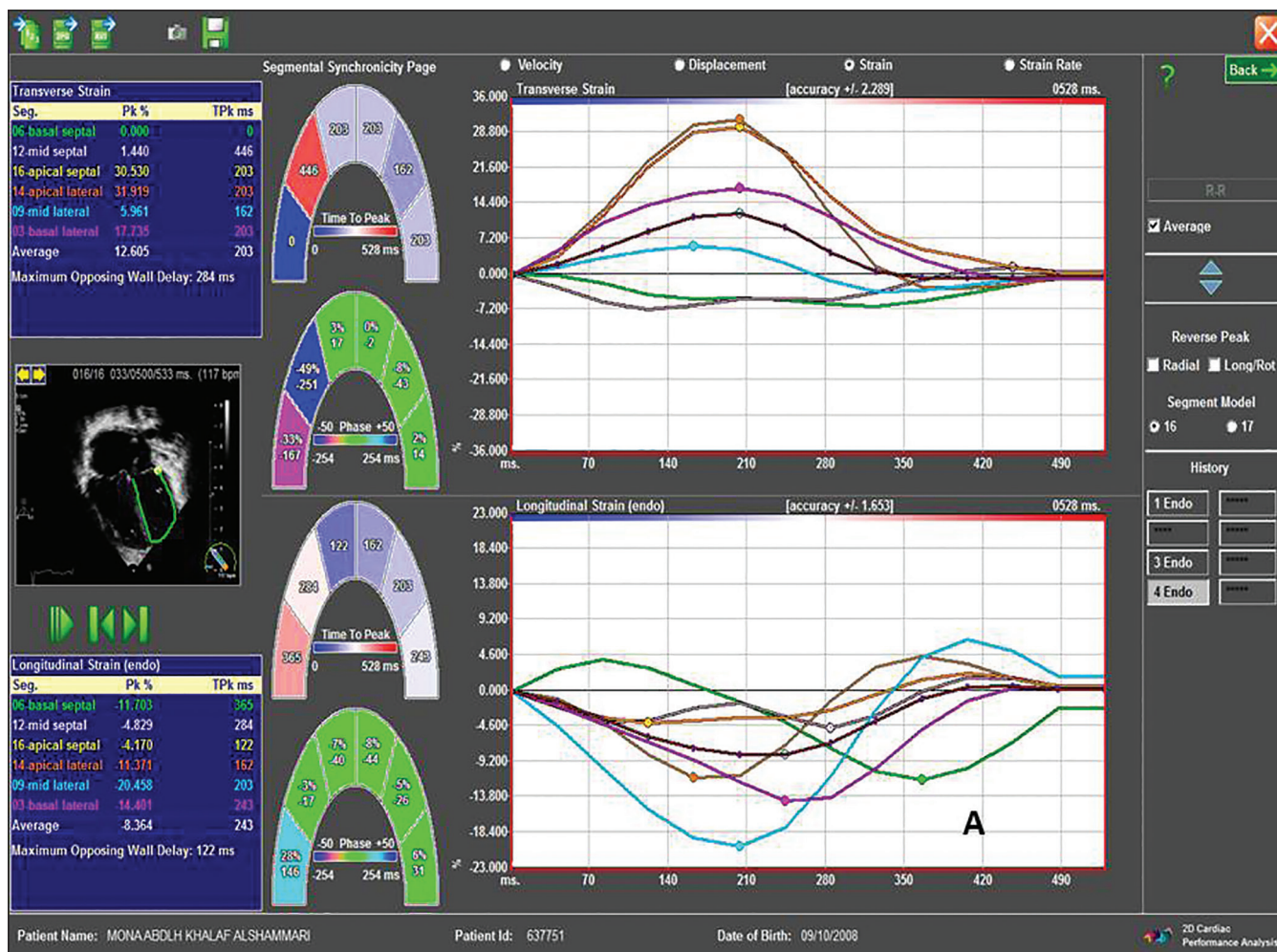


Figure 2: Speckle tracking analysis of the left ventricle (LV) from the apical four chamber view revealing a significant abnormally reduced LV deformation (A)

Previous studies already demonstrated that LVNC with preserved LVEF has subclinical myocardial dysfunction with the impairment of myocardial deformation parameters.^[6]

Myocardial fibrosis could probably explain the abnormal myocardial deformation properties found in our patient in the presence of a normal LV ejection fraction. Nucifora *et al.* demonstrated in about half of the patients with isolated LVNC, fibrosis involving both compacted and NC myocardium with a similar prevalence.^[7]

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Conflicts of interest

There are no conflicts of interest.

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