Dilated Cardiomyopathy in Friedreich’s Ataxia: 2D Echo and Tissue-Doppler Analysis of Left Ventricular and Atrial Function

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Introduction

Friedreich’s ataxia (FA) is a hereditary spinocerebellar degenerative disease characterised clinically by ataxia, dysarthria, skeletal deformities, and progressive dystrophia of the skeletal muscles. The disease is frequently associated with concentric and, in some cases, eccentric hypertrophic cardiomyopathy. Presentation of a dilated cardiomyopathy with global dysfunction of the myocardium is rare and commonly supposed to represent an end-stage of a progressive transition from the hypertrophic cardiomyopathy[1]. We report the echocardiographic and tissue-Doppler findings in a case with FA who presented with dilated cardiomyopathy shortly after debut of cardiac symptoms.

Case Report

A 25-year-old man with known FA was evaluated for dry cough and ortopnea for 2 months. Typically, ataxia, tremor, muscle weakness, and atrophy of the limbs were noted during examination. Electrocardiogram (ECG) showed sinus rhythm with right axis deviation [Fig. 1(A)]. Echocardiography [Fig. 1(B, C)] demonstrated a dilated, hypokinetic left ventricle with a diastolic dimension of 5.5 cm (body surface area of only 1.48 m², i.e. 3.7 cm/m²), a fractional shortening of 11%, and ejection fraction of 25%. No left ventricular hypertrophy was present. In the apex of the left ventricle, a large mobile thrombus was noted. Negligible functional mitral regurgitation was also present. The left atrium was enlarged with a diameter of 4.7 cm. The mitral inflow demonstrated restrictive left ventricular filling pattern with high E peak velocity, a deceleration time of only 64 ms, with a very diminished or absent A wave [Fig. 1(D)]. From the upper parasternal short axis, the proximal parts of the right and left coronary arteries were visualised, which appeared to be normal.

Tissue-Doppler examination was performed in the standard apical views with a GE Vingmed Vivid Five equipment and tissue-tracking presentation. This presentation visualises regional systolic shortening from base towards apex, depicted as coloured bands[2]. In the normal heart, myocardial shortening near the atrioventricular plane is above 12 mm, and tissue tracking shows that the longitudinal systolic shortening amplitude decreases gradually from base to apex [Fig. 2(A)]. The patient showed extremely reduced left ventricular systolic shortening from base to apex [Fig. 2(B)]. Tissue tracking during the PQ interval of the cardiac cycle shows apical motion of the posterior walls of the atria in the normal heart displaying atrial contraction [Fig. 2(C)]. However, in the FA patient, no mechanical left atrial systole could be detected either by tissue tracking from the apex [Fig. 2(D)] or by pulsed tissue-Doppler velocity from parasternal, apical [Fig. 2(D)], and subcostal views.

No embolism occurred during anticoagulant therapy, and the clinical condition improved on treatment with diuretics, digoxin, and increasing the
dosages of angiotensin-converting enzyme inhibitor and betablocker.

Discussion

The most common cardiac abnormality in FA is asymmetrical or concentric left ventricular hypertrophy associated with abnormal left ventricular diastolic filling, but with preserved systolic function in terms of a normal ejection fraction. Recently, a tissue-Doppler imaging study in FA patients without cardiac symptoms demonstrated decrease of systolic and early diastolic myocardial velocity gradients of the left ventricular posterior wall obtained from parasternal long-axis view[3]. This study indicates that left ventricular radial systolic function is likely to be decreased in these hypertrophic ventricles with normal ejection fraction. The present case indicates that heart involvement in FA may present as a dilated cardiomyopathy at the onset of symptoms, and the disease affects shortening of myocardial fibres with a circular (radial) orientation (fractional shortening) as well as fibres with a longitudinal course (tissue tracking). In addition, in FA cardiomyopathy, mechanical left atrial systole may be abolished despite sinus rhythm. As in other cases of severe left ventricular dysfunction, the condition may be complicated by apical thrombus formation. The genetic basis for the disease is a GAA trinucleotide repeat expansion in FA gene, which seems to lead to reduced levels of the protein, frataxin, resulting in abnormalities of mitochondrial iron and antioxidant systems. The abnormalities of left ventricular radial, longitudinal, and left atria systolic function in the present case might be related to a special genotype of FA. Accordingly, a significant association has been shown between the number of GAA repeats and left ventricular myocardial systolic and diastolic velocity gradients[3]. However, a relationship between the cardiac and genetic phenotype in the present case remains unknown as no genetic information is available.

Figure 1. A 12-lead ECG with sinus rhythm from the FA patient (A); M-mode curve of the left ventricle with end-diastolic and systolic dimensions (B); apical four-chamber view with apical mural thrombus (C); transmitral Doppler flow with absence of atria filling flow (D).
ability to measure both regional and global left ventricular radial and long-axis function, and supplements the information of myocardial characteristics given by conventional echocardiography. The method seems comparable in measuring radial and long-axis cardiac function with other imaging techniques such as three-dimensional tagged magnetic resonance imaging[4]. Tissue-Doppler imaging has the potential to detect subclinical heart disease and might be able to differentiate between different types of dilated cardiomyopathy, which must be addressed in future studies.

In conclusion, applying tissue-Doppler imaging in this case gave important additional information of the left atria and ventricular systolic function.

References