CLINICAL VIGNETTE

Nonroutine use of 2-dimensional speckle tracking echocardiography and fatigue assessment to monitor the effects of therapeutic venesections in a patient with newly diagnosed hereditary hemochromatosis

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Hereditary hemochromatosis (HH) is a genetic disease which, in over 80% of cases, is related to the C282Y mutation in the *HFE* gene. This condition leads to excessive iron absorption and accumulation in different organs, including the heart, and ultimately their damage. The process of cardiac involvement is typically slow and extended in time. Incremental accumulation of iron in cardiomyocytes induces oxidative stress and causes tissue damage which eventually results in heart failure, always associated with a bad prognosis.[1] Although therapy reducing overall iron level may reduce cardiac lesions, it is crucial to prevent their progress by the upfront treatment at the preclinical phase of the disease. However, during the early phase of the disease, when patients do not present severe abnormalities such as increased serum iron parameters and severe symptoms of impairment of other organs, heart damage may be even overlooked due to the lack of evident signs of cardiac dysfunction in the routine cardiac screening. The most important diagnostic tools to detect myocardial infiltration are cardiac magnetic resonance (CMR) and echocardiography; however, despite the indisputable value of CMR,[2] this is not a first-line diagnostic technique in the daily clinical practice. Echocardiography, as a widely available modality, with special attention to 2-dimensional speckle tracking echocardiography (2D STE), allows the detection of discrete abnormalities of the heart,[3] including patients with HH as we showed in our previous study.[4]

We present the case of a 42-year-old man with newly diagnosed HH (C282Y/H63D mutation in the *HFE* gene) with a nonroutine use of 2D STE and authorial fatigue assessment scheme in the monitoring of the effects of venesection treatment. Echocardiography was performed at the time of diagnosis and after 6 months from venesection therapy. The Fatigue Assessment Scale[5] (FAS; assesses the overall fatigue), the Chalder Fatigue Scale (CFQ; assesses fatigue in the last month), and the Fatigue Severity Scale (FSS; assesses fatigue in the last week) were carried out before and up to 2 weeks after each venesection.

During the 6-month follow-up, the patient had 6 venesections, achieving an overall reduction of ferritin level from 663 to 175 ng/ml (Supplementary material, *Table S1*). Echocardiography was performed using the GE VIVID E95 ultrasound system (GE Ultrasound, Horten, Norway) equipped with phased-array transducer (M5S). For each view, 3 consecutive cardiac cycles were recorded during quiet respiration; gray scale recordings were optimized for left ventricular (LV) evaluation at a frame rate of 50 to 80 frames/s; echocardiograms were stored digitally, and further offline analysis was performed using a commercial EchoPAC workstation (v201, GE Healthcare Horten, Norway). On

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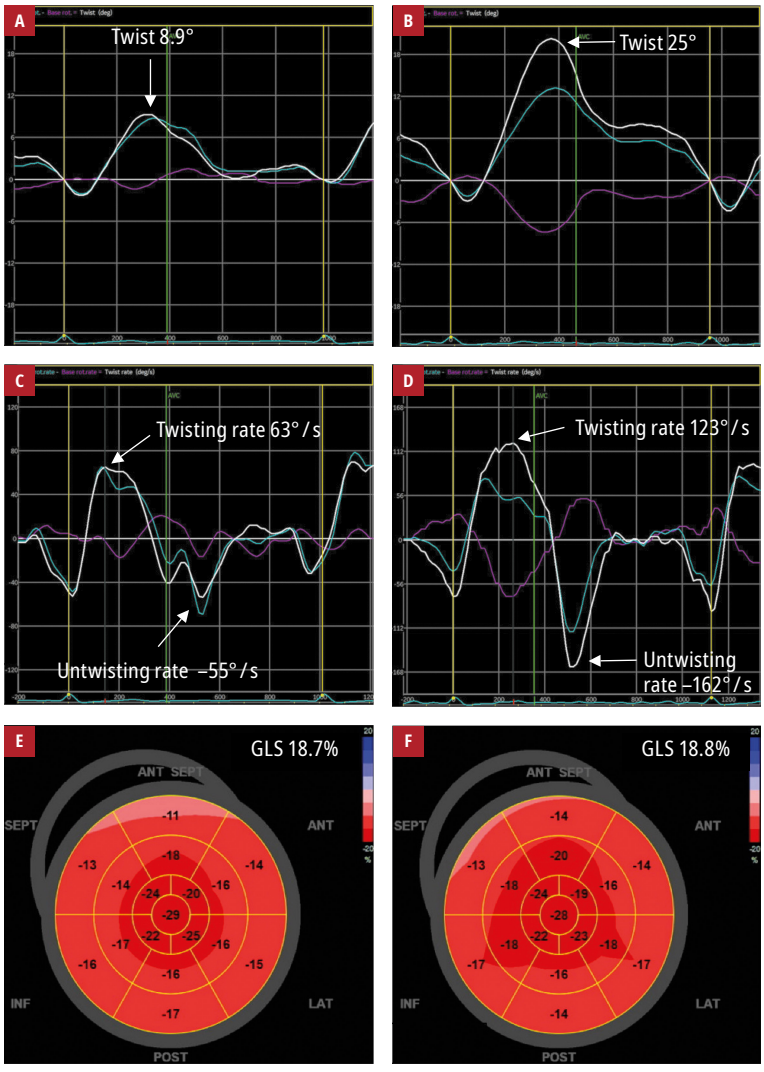

FIGURE 1 Two-dimensional speckle tracking echocardiography of a 42-year-old man with early diagnosed HH before (**A**, **C**, **E**) and after 6 months (**B**, **D**, **F**) of venesections. Twist (**A** and **B**), left ventricular twisting rate, and left ventricular untwisting rate (**C** and **D**) increased significantly; there were no changes in global longitudinal strain which was at a lower level of normal (**E** and **F**).

echocardiography, standard parameters were normal and did not change (Supplementary material, *Table S2*), while the parameters of rotation (twist, torsion, LV twisting rate, and LV untwisting rate) increased significantly; there were no changes in global longitudinal strain (GLS) which was at a lower level of normal (FIGURE 1A-1F). In the fatigue assessment, a gradual improvement was observed in all of the scales (in the FAS, reduction from 25 to 17 points, in the CFQ, from 12 to 5 points, in the FSS from 37 to 13 points) with the most spectacular effect after the 4th venesection (Supplementary material, *Table S3*).

Venesections significantly reduce the intensity of fatigue in patients with HH, which may be estimated with the aforementioned scales. The effects of this treatment in case of myocardial function can be monitored by speckle tracking echocardiography, which allows us to discover subtle changes in the heart, but this is not a standard procedure. It is difficult to connect the increase in LV rotation and the improvement in the quality of life in patients with near-normal resting echocardiography (except lower rotation and borderline GLS); however, rotation abnormalities could be symptomatic and, when corrected, paralleled by clinical improvement. All conclusions listed above require further observation in appropriate groups of patients with HH, including hemodynamic parameters, such as natriuretic peptides.

SUPPLEMENTARY MATERIAL

Supplementary material is available at www.mp.pl/kardiologiapolska.

ARTICLE INFORMATION

CONFLICT OF INTEREST None declared.

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