A 47-year-old man presented with 1-week history of cough and bilateral lower extremity swelling. He had a 5-year history of diabetes mellitus and hypogonadism. His ECG demonstrated sinus rhythm with low-limb voltage and left anterior hemiblock. An echocardiogram showed severely reduced left ventricular ejection fraction 10%, restrictive diastolic filling, and severe right ventricular dysfunction (Figure 1, Movie I–IV in the online-only Data Supplement). Laboratory testing revealed brain natriuretic peptide 1849 pg/mL, ferritin 6389 µg/L, iron 108 µg/dL, transferrin 124 mg/dL, total iron binding capacity 171 µg/dL, and transferrin saturation 63%.

Cardiac catheterization demonstrated angiographically normal coronary arteries, heart rate 87 beats per minute, blood pressure 104/74 mm Hg, right atrial pressure 24 mm Hg, right ventricular pressure 37/24 mm Hg, pulmonary artery pressure 37/25 mm Hg, and mean pulmonary capillary wedge (PCW) pressure 25 mm Hg (Figure 2). The cardiac index was 1.7 L/min per m² with a systemic vascular resistance of 1411 dynes/cm² and a pulmonary vascular resistance of 94 dynes/cm². Endomyocardial biopsy confirmed extensive iron deposition in myocytes (Figure 3). A liver biopsy demonstrated stage 3 to 4 fibrosis and elevated iron content. Genetic testing confirmed homozygous HFE C282Y mutation.

Symptomatic hypotension limited medical therapy to torsemide, spironolactone, and digoxin. Phlebotomy of 250 mL, 3 times a week, and desferasirox resulted in decreased serum ferritin to 53 µg/L within 9 months with improved functional status from New York Heart Association class IV to I. Repeat echocardiogram showed marked improvement of left ventricular function (Figure 1, Movie I–IV in the online-only Data Supplement).
ejection fraction to 45% (Movie V–VIII in the online-only Data Supplement) and a brain natriuretic peptide of 7 pg/mL.

Systolic left ventricular dysfunction in hereditary hemochromatosis portends a poor prognosis despite reports of clinical improvement with phlebotomy since the 1950s.1,2

In general, weekly removal of 400 to 500 mL of blood is recommended. In hemodynamically tenuous patients, more frequent but small volume phlebotomy may be successfully performed. Iron-chelation therapy may also facilitate iron removal even in primary hemochromatosis.3

Although standard systolic heart failure therapy with β-blockers, angiotensin-converting enzyme inhibitors, and aldosterone antagonists may be limited by blood pressure in such patients, hypotension should not interfere with the definitive therapy of phlebotomy. In this case, the improvement in cardiac function was almost entirely attributable to iron removal because standard heart failure therapy was limited by hypotension.

Disclosures

None.

References


Key Word: nonischemic cardiomyopathy

Figure 2. Elevated right atrial (RA) and pulmonary capillary wedge (PCW) pressures with prominent Y descents (arrows, A and D) and dip-and-plateau right ventricular (RV) diastolic pressure tracing (arrows, B) is consistent with restrictive ventricular filling. C, Mild pulmonary hypertension is observed. PA indicates pulmonary artery.

Figure 3. A, Iron stain, 400X: Extensive iron deposition within cardiac myocytes. B, Iron stain of hepatocyte, 100X: Significant hepatocellular iron deposition.
Reversal of Severe Biventricular Dysfunction From Cardiac Hemochromatosis With Iron Removal
Chen H. Chow, Chantal El-Amm, Wendy Liu, Stephen Pastva, Ilke Sipahi and James C. Fang

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