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Rare case of Fabry disease cardiomyopathy and severe mitral stenosis

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A 44-year-old woman was admitted to hospital for diagnostics. Undergoing dialysis therapy for approximately 10 years. Transthoracic echocardiography revealed a preserved left ventricular ejection fraction (60%) and severe mitral stenosis (MS) with minor regurgitation. Concentric hypertrophy of the left ventricle (with an interventricular septum of 22 mm) was noted (Figure 1A), with an intraventricular gradient — 73 mm Hg. Transesophageal echocardiography confirmed the severity of the MS. The patient was considered a candidate for valvular surgery in the event of rapid clinical deterioration. Coronary angiography was performed on this patient, and no significant obstructions were found. Cardiac magnetic resonance imaging showed diffuse, non-ischemic myocardial fibrosis in the basal and middle segments of the posterior wall, as well as the basal segment of the inferior and interventricular septum (Figure 1B–C). The image was inconclusive. Speckle tracking echocardiography showed hypokinesis of the basal segment of the inferior and posterolateral wall, with a global

longitudinal strain of –12.3% (Figure 1D). Anderson–Fabry disease was suspected, and subsequently confirmed by testing for α-galactosidase A deficiency. Genetic testing also confirmed the diagnosis of Fabry disease, with a heterozygous mutation detected: c.1025G>A (p.Arg342Gln).

The patient was referred for enzyme replacement therapy, using agalsidase beta, at a tertiary care center. Two of her sons were also diagnosed with this condition, both of whom have since started early treatment. It is hoped that this early pharmacological intervention will help them avoid the long-term consequences of the disease.

A follow-up visit showed a decrease of left ventricular wall thickness (interventricular septum of 19 mm). The patient reported a significant increase in exercise capacity. Moreover an improvement in global longitudinal strain (–15.8), could also be measured. (Figure 1E).

Anderson–Fabry disease is an uncommon etiology of left ventricular hypertrophy, with valvular involvement being particularly rare.[1, 2] To our knowledge, this is the first case of severe MS in a patient with Fabry disease reported in the literature. Speckle tracking echocardiography proves to be a valuable tool in the differential diagnosis of cardiac hypertrophy, which should be considered especially in patients where this finding is accompanied by renal disease, as nephropathy is one of the major complications of Fabry disease.

Article information

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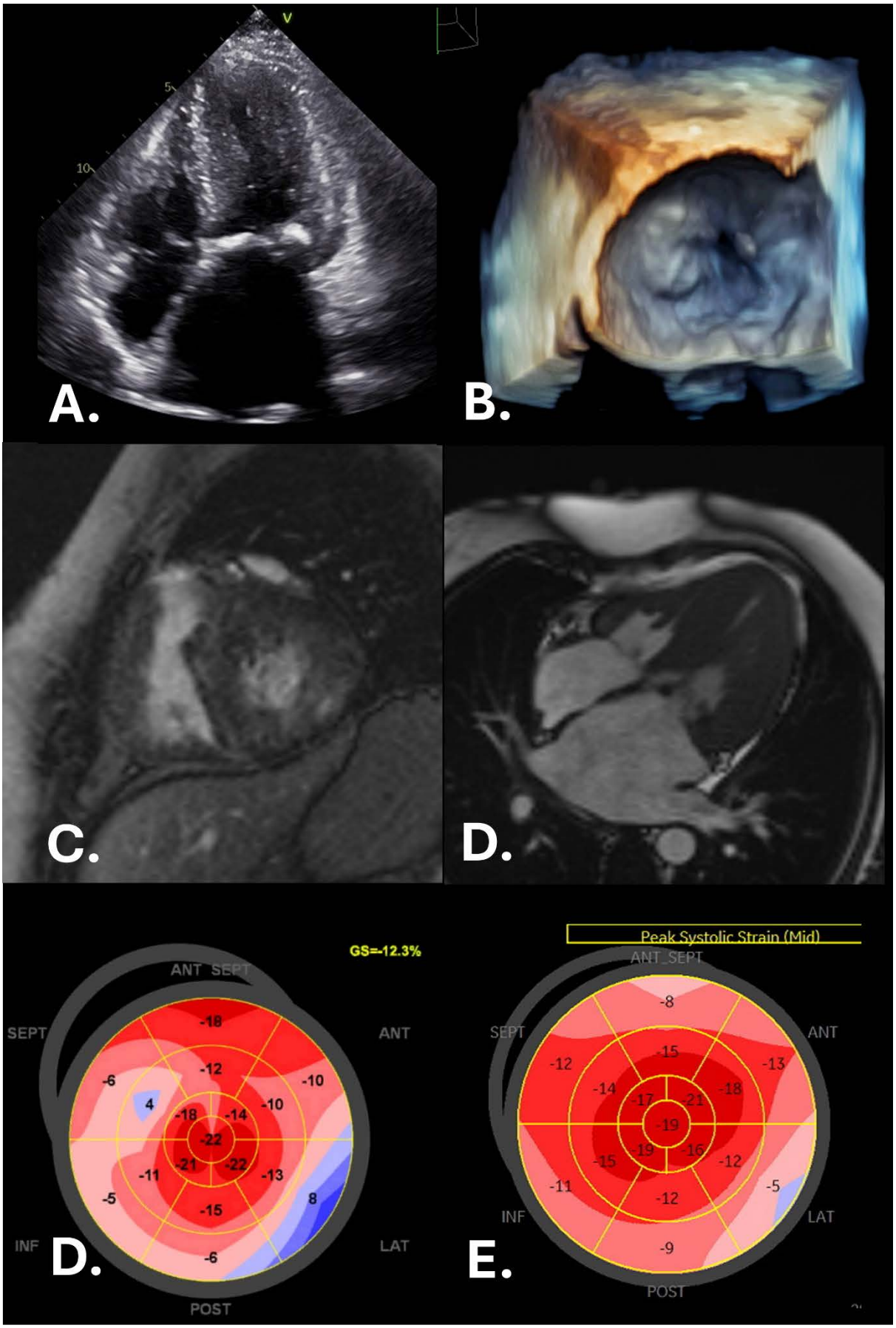
A.
B.
C.
D.
GS=-12.3%
ANT SEPT
SEPT
ANT
INF
LAT
POST
-18
-12
-6
-10
4
-18
-14
-10
-22
-21
-22
-11
-13
-5
-15
8
-6
D.
Peak Systolic Strain (Mid)
ANT_SEPT
SEPT
ANT
INF
LAT
POST
-8
-15
-12
-13
-14
-17
-21
-18
-19
-15
-19
-16
-12
-11
-12
-5
-9
E.

Figure 1. A. Four-chamber view in transthoracic echocardiography showing left ventricular hypertrophy. **B.** 3D Echocardiography image showing a heavily calcified mitral valve, which caused severe mitral stenosis **C.** Short-axis view of cardiac magnetic resonance shows diffuse, non-ischemic myocardial fibrosis. **D.** Cardiac magnetic resonance 4-chamber view showing left ventricular hypertrophy during systole. **E.** "Bulls-eye" speckle echocardiography pattern showing hypokinesis of the basal segment of the inferior and posterolateral wall. **F.** "Bulls-eye" speckle echocardiography pattern showing improvement of left ventricular function measured by global longitudinal strain after enzyme replacement therapy