Apical Sparing of Longitudinal Strain, Left Ventricular Rotational Abnormalities, and Short-Axis Dysfunction in Primary Hyperoxaluria Type 1

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A 24-year-old woman with end-stage renal failure because of primary hyperoxaluria type 1 was evaluated in our hospital for systemic calcium oxalate deposition in the course of long-term (5 years) hemodialysis therapy. Diagnosis of primary hyperoxaluria type 1, a hereditary cause of calcium oxalate kidney stones or progressive nephrocalcinosis that frequently results in end-stage renal failure, was made by liver biopsy (reduced alanine:glyoxylate aminotransferase activity, 3.0 µmol/h per milligram protein; normal, 19.1–47.9) and by genetic testing (homozygosity for the c.302 T>C, AGXT mutation). Her plasma oxalate level on regular hemodialysis (3× per week over 4 hours) was increased (86 µmol/L predialysis; normal, <10 µmol/L).

Transcatheter echocardiography revealed increased wall thickness of the left ventricle (LV; Figure 1). The LV was mildly dilated and showed a decreased systolic function. Flow across the mitral valve demonstrated a restrictive filling pattern. Moreover, the myocardium had a characteristic echo-dense granular sparkling appearance (Movie in the online-only Data Supplement). Both atria were enlarged. Moderate tricuspid regurgitation was present. A right ventricle systolic pressure of 62 mm Hg was calculated.

We used 2-dimensional speckle tracking echocardiography to assess the multidirectional myocardial function. Global LV longitudinal peak systolic strain (LS) was impaired (−13.5%; normal, −20.9±1.3%). LS was severely reduced in the basal and midventricular segments of the lateral and septal LV walls. Moreover, LS in the apical segments was relatively preserved resulting in a typical apical sparing strain pattern (Figure 2). Short-axis myocardial function was also affected (Figure 3), and reversed LV rotation at the basal level with loss of LV twist mechanics was noted, suggesting impairment of both systolic and diastolic myocardial LV function (Figure 4).

Currently, the patient experiences increasing oxalate osteopathy and heart failure. We, hence, recommended to increase the hemodialysis regimen to 6×3–4 hours/week to achieve a sufficient removal of the body oxalate stores and hence, she would urgently need combined or sequential liver–kidney transplantation.

Discussion

This report highlights the adjunctive role of 2-dimensional speckle tracking echocardiography in metabolic conditions such as primary hyperoxaluria type 1. Although typical characteristics of infiltrative cardiomyopathy with restrictive physiology were easily recognized using conventional echocardiography in our patient, 2-dimensional speckle tracking echocardiography identified important additional echocardiographic features, which may increase our understanding of the mechanisms contributing to cardiac dysfunction in systemic oxalosis.

Apical sparing is a pattern of regional differences in deformation, in which LS in the basal and midsegments of the LV is more severely impaired compared with the strain values in the apical segments. A potential pathophysiologic mechanism of apical sparing may be the heterogeneous myocardial deposition of oxalate crystals. In our patient, wall thickness was mainly increased in the basal and midventricular segments compared with the modestly increased wall thickness at the apex. This may indicate that less oxalate crystals accumulated in the apex, resulting in the apical sparing LS pattern and relatively preserved apical rotation mechanics.

Phelan et al4 were the first to demonstrate the clinical relevance of this strain pattern in patients with cardiac amyloidosis. In their study, apical sparing differentiated cardiac amyloidosis from other causes of LV hypertrophy with a high sensitivity and specificity.4 To the best of our knowledge, we report the first case of apical sparing in primary hyperoxaluria type 1 associated with infiltrative cardiac disease. Therefore, we suggest that apical sparing may be a typical diagnostic feature of infiltrative cardiomyopathies in patients with a systemic disease characterized by myocardial accumulation of toxic substances, such as amyloid and oxalate crystals.
Disclosures
None.

References

Key Words: cardiomyopathy, restrictive ▪ echocardiography ▪ hyperoxaluria, primary ▪ oxalosis

Figure 1. Apical 4-chamber view demonstrating increased wall thickness of the septal (purple arrow) and lateral (green arrow) left ventricle walls. The wall thickness at the apex was only modestly increased (yellow arrow).

Figure 2. Left ventricular longitudinal peak systolic strain (LS) was assessed using standard apical views (automatic function imaging, GE Healthcare, Horten, Norway). A bull’s-eye plot showing global and regional LS was automatically generated. Note the apical sparing pattern of LS, which is explained by regional differences in LS between the apical, midventricular and basal segments. The apical sparing pattern was quantified by expressing the average apical LS (−19.4%) as a quotient of the sum of the average mid (−11.3%), and basal (−5.3%) LS values, as previously described. This quotient was defined as relative apical LS. The relative apical strain in our patient was 1.16. A cutoff value of 1.0 was used in the study by Phelan et al to differentiate cardiac amyloidosis from other causes of LV hypertrophy.
Figure 3. Short-axis dysfunction. Time-strain color-coded curves were obtained from midventricular short-axis views. Systolic peak radial (A) and circumferential (B) strain was measured in each of 6 LV segments (yellow, anteroseptal; light blue, anterior; green, lateral; purple, posterior; dark blue, inferior; and red, septal). Global (average of 6 segments) radial (A) and circumferential (B) peak systolic strain values were reduced (+27.1%; normal, +57.3±5.0% and −16.7%; normal, −21.8±1.5%, respectively), particularly in the septal, anteroseptal, and inferior segments.

Figure 4. Abnormal left ventricular twist mechanics. In our patient, basal rotation (purple curve) was reversed (counterclockwise, positive value) with subsequent loss of normal twist (white curve). Left ventricular rotation was preserved at the apex.
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