

Subtle cardiac dysfunction in nephropathic cystinosis: insight from tissue Doppler imaging and 2D speckle tracking echocardiography

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Abstract

Background

Nephropathic cystinosis is a rare autosomal recessive lysosomal storage disorder that initially affects the kidney progressing to multi-organ failure due to accumulation of cystine in all tissue compartments.

Objective

The main objective of this study is the evaluation of cardiac function in cystinosis patients using non-conventional echocardiographic modalities like pulsed wave tissue Doppler imaging (PW-TDI) and 2D speckle tracking echocardiography (2D-STE).

Methods

This is a case control study conducted on fifteen patients with cystinosis and 15 normal controls. Echocardiography was done for all participants and PW-TDI was performed for measurement of S' , E' , A' velocities and myocardial performance index (MPI) at basal parts of septal, left ventricle (LV), and right ventricle (RV) free walls. 2D-STE was done for evaluation of global longitudinal strain (GLS), global circumferential strain (GCS), and global radial strain (GRS) of LV. Mitral E and A velocities and tricuspid annular plane systolic excursion (TAPSE) were also measured.

Results

The GLS, GRS, and S' velocity at basal septum and LV lateral wall were significantly lower in patients denoting LV systolic dysfunction ($p = 0.005$, $p < 0.0001$, $p = 0.001$, $p = 0.006$, respectively), while E/E' were significantly higher in patients group denoting LV diastolic dysfunction ($p < 0.001$). For RV function, TAPSE, S' , and E' velocity were significantly lower in patients group ($p = 0.013$, $p < 0.01$, $p = 0.05$,

respectively) indicating RV systolic and diastolic dysfunction. The TDI-derived MPI for both LV and RV were significantly higher in patients group ($p < 0.0001$, $p < 0.01$, respectively) indicating both ventricular systolic and diastolic dysfunction. For prediction of cardiac dysfunction among patients, the receiver operating characteristic (ROC) curve showed that $GRS \leq 29\%$ had sensitivity 93.3% and specificity 100%, $GLS > -20.1\%$ had sensitivity 66.7% and specificity 93.3%, $LV-E/E' > 7.87$ had sensitivity 73.3% and specificity 93.3%, and $MPI-LV > 0.36$ had sensitivity 100% and specificity 93.3% while $MPI-RV > 0.29$ had sensitivity 80% and specificity 93.3% and $TAPSE \leq 19$ mm had sensitivity 80% and specificity 73.3%.

Conclusions

Patients with cystinosis have significant both left and right ventricular dysfunction, which can be better evaluated using the non-conventional echocardiographic modalities like TDI and 2D-STE for early detection of subtle cardiac dysfunction.

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