New echocardiographic approaches to differentiate cardiac amyloidosis and Fabry disease: the right heart and mitral valve thickness

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Background: Infiltrative cardiomyopathies such as cardiac amyloidosis (CA) and Fabry disease (FD) are associated with high cardiovascular morbidity and mortality. Diagnosis is often challenging as CA and FD may present similar cardiac phenotypes on standard echocardiographic imaging, especially in early stages of disease.

Purpose: Our study focuses on right ventricular (RV) two-dimensional speckle tracking imaging and mitral valve thickness as diagnostic strategies to discriminate CA from FD. Here, we report preliminary data of our study registry of CA and FD patients.

Methods: We retrospectively screened patients with diagnosed CA or with genetically confirmed FD from our study registry. Global and free wall RV strain and mitral leaflet thickening in both groups were analysed using 2D speckle tracking echocardiography and linear measurements from a parasternal long axis view, respectively (Vivid E9 or E95, GE Vingmed, Horten, Norway, M5S 1.5–4.5MHz transducer). The diagnostic accuracy for discriminating CA and FD in both groups was evaluated using receiver operating characteristic (ROC) curve analysis.

Results: A total of 33 FD and 27 CA patients, including 17 patients with wild-type transthyretin-related (ATTR) CA, 8 patients with hereditary ATTR CA, one patient with light chain CA, and one patient with AA CA, were included. RV function was significantly reduced in CA compared to FD patients as measured by global longitudinal RV strain (−13.6±5.1 in CA vs. −18.4±3.9 in FD, p<0.001) and free wall longitudinal RV strain (−17.5±5.7 in CA vs. −21.3±4.4 in FD, p=0.005). Significantly lower longitudinal RV strain was observed in the basal to mid ventricular region in CA compared to FD patients, while RV apical strain was not significantly different between the two groups. RV global longitudinal strain had the highest diagnostic accuracy of strain analyses (area under the curve [AUC] 0.79, 95% confidence interval [CI] 0.66–0.93) to discriminate CA from FD. Additional measurement of the mitral valve in mid diastole in the parasternal long axis view revealed a significantly thickened leaflet in CA patients (4.6±1.2 mm in CA vs. 2.9±1.0 mm in FD, p<0.001) corresponding to an AUC of 0.84 (95% CI 0.72–0.96) in ROC analysis.

Conclusion: Global longitudinal RV strain was significantly reduced in CA patients while the mitral valve leaflets were significantly thickened compared to FD patients. Therefore, both parameters may be useful to discriminate CA from FD in the echocardiographic workup of patients with unclear left ventricular wall thickening.