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Cardiac amyloidosis mimicking hypertrophic obstructive cardiomyopathy: role of multimodality imaging and genetic testing

S. Kumar1, M. Yadava2, S. Heitner2

(1) University of Hawaii, Honolulu, United States of America

(2) Oregon Health & Science University, Portland, United States of America

Abstract

Cardiac amyloidosis is an infiltrative cardiomyopathy characterized by concentric ventricular hypertrophy. Asymmetric septal hypertrophy is a rare finding that has only been reported in a handful of cases. We describe a case of cardiac amyloidosis mimicking hypertrophic obstructive cardiomyopathy (HOCM) in which multimodality imaging helped identify the correct diagnosis.

Case: A 65 year-old man was noted to have an abnormal EKG showing left bundle branch block and first degree atrioventricular block. Echocardiography demonstrated asymmetric septal hypertrophy, systolic anterior motion of the anterior mitral valve leaflet, and a peak left ventricular outflow tract gradient of 38 mmHg at rest. Based on these findings a presumptive diagnosis of HOCM was made. Cardiac magnetic resonance confirmed the asymmetric septal hypertrophy with a maximal wall thickness of 24 mm. There was evidence of late gadolinium enhancement in the proximal interventricular septum, bilateral atria and right ventricle free wall. Strain echocardiography demonstrated relative apical sparing pattern, an unusual finding for HOCM. Given the atypical findings for HOCM, an endomyocardial biopsy was undertaken which showed evidence of amyloidosis. Subsequently genetic testing revealed a Val30Met mutation in the TTR gene confirming transthyretin-related hereditary amyloidosis.

Biography:

Suwen Kumar has completed his medical school from All India Institute of Medical Sciences, New Delhi, India. After finishing residency in Internal Medicine from Michigan State University, he did Advanced Cardiac Imaging Fellowship from Oregon Health & Science University before joining University of Hawaii for Cardiology Fellowship.

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