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CARDIOVASCULAR FLASHLIGHT
Cardiac imaging

Pseudoxanthoma elasticum causing extensive myocardial calcification

Elliott J. Carande *, Elliw F. Roberts, Abigail M. Probert, Benjamin Dicken, and Adrian Ionescu

Morriston Hospital, Swansea Bay University Health Board, Swansea SA6 6NL, UK

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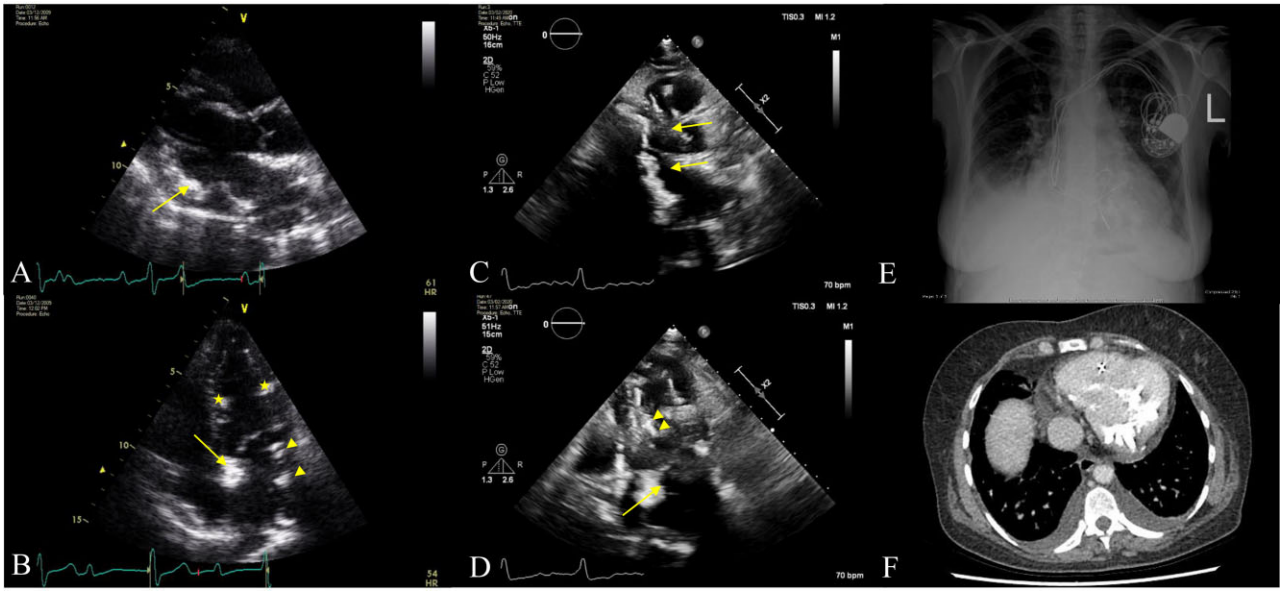

A 51-year-old lady with pseudoxanthoma elasticum and bilateral retinal angioid streaks, presented with generalized tonic-clonic seizures. Medical history included a permanent pacemaker for complete heart block, and permanent atrial fibrillation (AF). Due to an incidental finding of a distended abdomen with deranged liver function tests, an abdominal ultrasound was performed, which demonstrated gross ascites and a cirrhotic-looking liver. Given findings of bilateral pleural effusions and pulmonary oedema, and elevated NT pro-BNP (14000 ng/L), a transthoracic echocardiogram was performed.

Transthoracic echocardiogram demonstrated extensive myocardial calcification in both ventricles with an estimated left ventricular ejection fraction of 40–50%, markedly worse than 12 years previously (*Panels A–D* and Supplementary material online, *Videos S1* and *S2*). The mitral flow pattern in diastole was unimodal, in the context of paced rhythm and AF, with a short deceleration time of 144 ms and a pressure half-time of 34 ms, indicating restrictive physiology. Chest X-ray demonstrated calcific opacities over the heart silhouette (*Panel E*). Retrospective analysis of computed tomography imaging of the abdomen and pelvis performed 18 months previously for

* Corresponding author. Tel: +44 7784050930, Email: e.carande@gmail.com
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abdominal pain demonstrated extensive myocardial calcification in the left ventricular (LV) apex, which was incidentally encompassed in the field of view (*Panel F*). Cardiovascular magnetic resonance imaging was not performed.

Pseudoxanthoma elasticum is a rare, genetic disorder associated with mutations of the *ABCC6* gene on chromosome 16, typically causing an accumulation of calcium in connective tissue, and angioid streaks in the retina. Although cardiovascular complications occur mainly due to accelerated atherosclerosis, restrictive cardiomyopathy secondary to severe, diffuse myocardial calcification may occur and progress. Our patient also had conduction tissue disease requiring a pacemaker at the age of 39, probably another manifestation of cardiac calcification and fibrosis. Treatment options for the restrictive cardiomyopathy are limited to diuretics, and the outlook is dismal.

Panel: (*A*) PLAX 2009—patchy calcification of the basal and mid-inferolateral segments (arrow). (*B*) Apical 4-chamber 2009—calcification of the crux of the heart (arrow) and the lateral left atrial wall and mitral annulus (arrowheads) as well as focal calcification of the left ventricular myocardium (asterisks). (*C*) PLAX 2021—extensive intra-cardiac calcification (arrows) completely obscuring the left ventricle. (*D*) Apical 4-chamber 2021—extensive progression of the calcification at the crux (arrow) and in the myocardium (arrowheads). (*E*) Chest X-ray from admission demonstrated radio-opaque features across the myocardium. (*F*) Extensive myocardial calcification was visible on the computed tomography scan.

Supplementary material

Supplementary material is available at *European Heart Journal – Case Reports* online.

Consent: The authors confirm that written consent for submission and publication of this case report including images and associated text has been obtained from the patient in line with COPE guidance.

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