Cardiac fibro-fatty masses in tuberous sclerosis: role of cardiovascular magnetic resonance

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A 19-year-old male with tuberous sclerosis (TS) was referred to the outpatient clinic of cardiology for the follow-up of cardiac rhabdomyomas. The diagnosis of TS was established in childhood based on the presence of large biventricular cardiac rhabdomyomas, epilepsy, and mental retardation. Magnetic resonance imaging of his brain showed tuberomas, hamartomas, and subependymal nodules. There was no involvement of other organs. The patient was asymptomatic and his examination was unremarkable. His ECG and Holter were normal. The echocardiogram showed mild dilatation of left cavities, normal biventricular function and hyperechoic areas in the septum, the anteroseptal, and the anterolateral walls of the left ventricle (Panels A–C).

The cardiovascular magnetic resonance (CMR) confirmed mild dilatation of the left cavities and revealed up to seven intramyocardial masses, the larger in the apex of the left ventricle (35 × 30 × 10 mm), with imaging features compatible with fibro-fatty tissue (Panels D–I).

Although rhabdomyomas are the typical cardiac finding in children with TS, the pattern of disease in adulthood is less known. The findings of this case suggest that rhabdomyomas can evolve to fibro-fatty tissue in the long term. An alternative hypothesis is that these lesions could be multifocal lipomas. However, the typical lipoma is not intramyocardial and does not have a strong component of late gadolinium enhancement (LGE).

This clinical case highlights the different roles of echocardiography and CMR in the study of cardiac masses in patients with TS, where CMR provides invaluable information about the number, location, and tissue characteristics of the lesions, helping in the management of the patient.

The two-dimensional echocardiogram revealed hyperechoic areas in the medial and apical segments of the interventricular septum (Panel A, arrow—parasternal long-axis view; Panel C, arrow-apical four-chamber view) and in the anteroseptal and anterolateral walls of the left ventricle (Panel B, arrow—parasternal short-axis view). On CMR the lesions revealed high signal at the b-steady-state free precession cine sequence (arrows in Panel D—four-chamber view; Panel E—two-chamber view; and Panel F—short-axis view) with a chemical shift artefact in the fat—water boundary; enhancement in the LGE sequence (Panel G—four-chamber view); high signal in T1 black-blood TSE (Panel H—four-chamber view); and low signal in the T1 black-blood TSE with fat supression (Panel I—four-chamber view). b-SSFP, steady-state free precession; TSE, turbo spin echo.

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