Arrhythmogenic Left Ventricular Cardiomyopathy Diagnosis: Benefiting from Advanced Cardiac Imaging

Diagnóstico de Cardiomiopatia Arritmogénica do Ventrículo Esquerdo por Imagem Cardíaca Multimodal

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We present the clinical case of a 70-year-old male with a history of hypertension, managed pharmacologically, presenting to the emergency department following an episode of syncope. This occurred suddenly without preceding symptoms and was followed by spontaneous recovery, despite generalized malaise and dizziness. A similar episode was reported on the previous year, with no further investigation. Upon admission, the patient exhibited tachycardia with a thready pulse and low blood pressure. Electrocardiography (ECG) depicted wide-complex tachycardia at 210 bpm, with a superior axis and a right bundle branch block pattern (Fig. 1A). Intravenous amiodarone was administered, resulting in conversion to sinus rhythm. Post-conversion ECG showed no significant abnormalities except for T-wave flattening with terminal inversion in leads V5-V6. Laboratory workup was largely unremarkable, except for elevated troponin and brain natriuretic peptide levels (BNP) (90 µg/L and 1200 pg/dL, respectively). Transthoracic echocardiography revealed a left ventricular ejection fraction (LVEF) of 40%, with suspected basal and mid inferolateral hypokinesia, mildly reduced global longitudinal strain (-16%) (Fig. 1B).

Further investigation with cardiac computed tomography angiography (CTA) was performed to exclude coronary artery disease. The coronary calcium score was 36 Agatston units, and no obstructive coronary disease was identified (CAD-RADS: 1/P1) (Fig. 1C, D, E). However, extracoronary findings were notable for reduced thickness of the basal and mid inferolateral walls, and the mid-lateral wall, with small areas within the myocardium of the lateral wall exhibiting reduced density (-50HU), in favour of the presence of fat (Fig. 2A, B, C). Cardiac magnetic resonance (CMR) imaging further elucidated the condition, revealing an LVEF of 38% with a non-dilated left ventricle, hypokinesia of the basal and mid inferolateral segments, and akinesia of the mid-lateral wall (Fig. 2D, E, F). The right ventricle was non-dilated

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with preserved function and no segmental abnormalities. Tissue characterization confirmed the presence of focal intramyocardial fat (evidenced by signal loss on T1-weighted fat-suppression sequences). Extensive subepicardial late gadolinium enhancement was noted in the basal and mid inferolateral and mid-lateral segments (Fig. 2G, H, I). Overall clinical presentation and imaging findings favoured the diagnosis of arrhythmogenic left ventricular cardiomyopathy (ALVC). The patient was started on guideline-directed medical therapy. Given the risk of sudden cardiac death, an implantable cardioverter-defibrillator was placed for secondary prevention.[1] Genetic testing was positive for c.2393_2394del, p.(Arg2326*) in plakophilin *PKP2* gene, and c.6976C>T, p.(Thr7981Ilefs*28) in filamin *C FLNC* gene, both in heterozygosity.

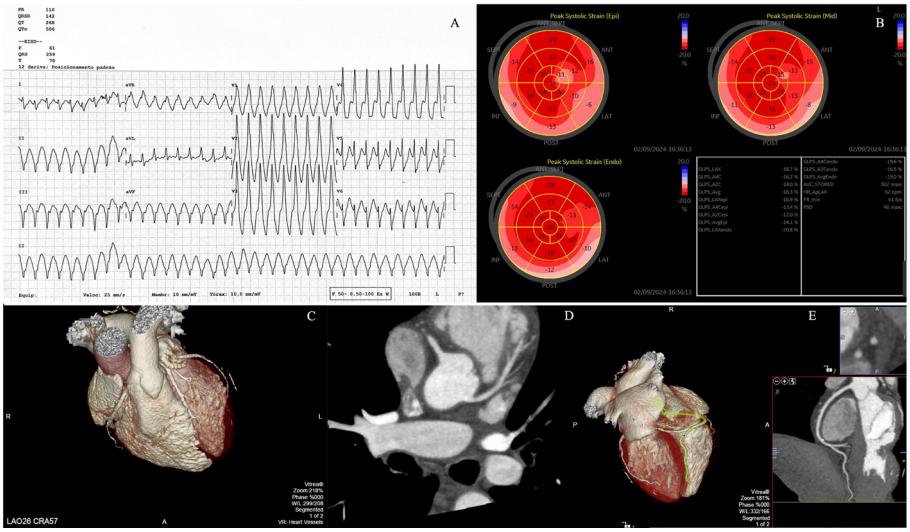

Figure 1. A – wide-complex right bundle branch tachycardia at the emergency department; superior oriented axis; B – Mildly reduced global longitudinal strain. Contrary to coronary artery disease, there is no specific regional longitudinal strain impairment. However, layer specific assessment, namely at the epicardial strain analysis, revealed more evident longitudinal strain affection, which stands for the diagnosis of an non-ischemic cardiomyopathy. C – volume rendering cardiac computed tomography (CT) image; D – CT axial views of the coronary arteries; E – curved planar reconstruction of the right coronary artery. There was non-obstructive coronary artery disease.

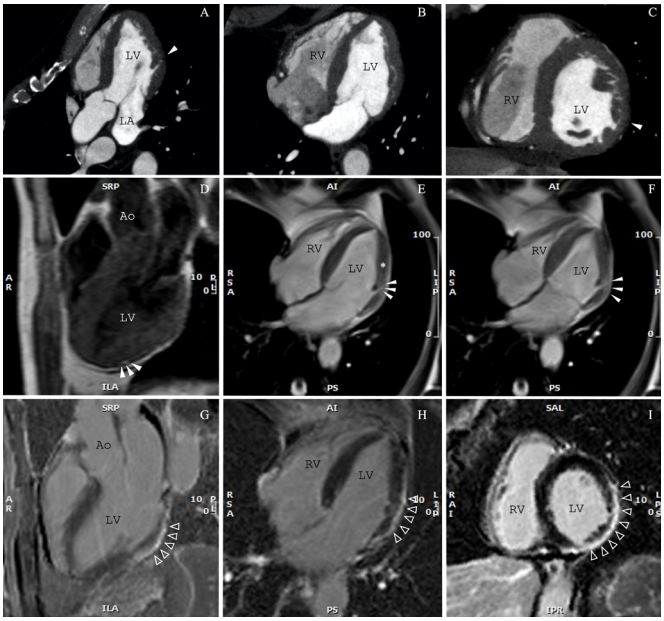

Figure 2. A-C: three-, four- and short axis cardiac views at cardiac CT. There is reduced LV wall thickness at the lateral and infero-lateral wall (arrow-head) with small areas of reduced/negative density within the myocardium, which is in favour of the presence of fat tissue. D – T1-weighted turbo spin Echo sequence at CMR with areas of fat infiltration (arrowheads) on the infero-lateral wall of the LV. E-F – CMR horizontal four-chamber view at cine (SSFP) images with mid-lateral LV wall akinesia (arrowheads) and intramyocardial chemical shift artifact (*), also favouring the presence of fat infiltration. G-I: extensive subepicardial late gadolinium enhancement at three, four-chamber and short-axis CMR views (arrowheads). LV – left ventricle; RV – right ventricle; LA – left atrium; Ao – Aorta; CMR – cardiac magnetic resonance; SSFP – steady-state free precession.

We found this case notable for the importance of clinical suspicion and advanced cardiac imaging workup in patients with unexplained syncope and ventricular arrythmias: 1) twelve-lead ECG revealed non-specific changes, contrary to what is described for classical right ventricular cardiomyopathy; 2) transthoracic echocardiography was an upfront diagnostic test albeit lacking myocardial tissue characterization; 3) cardiac CT excluded significant coronary artery disease as a more common aetiology and provided additional clues toward myocardial tissue changes; 4) multiparametric CMR was key for differential diagnosis, morpho functional assessment and detailed tissue characterization. Combined sequential use of distinct cardiac imaging modalities was critical for appropriate management and sudden cardiac death prevention.

ALVC is a recently described genetic cardiomyopathy characterized by the presence of myocardial fat and fibrosis, typically in a non-dilated LV. Advanced cardiac imaging, mostly settled on multiparametric CMR, is essential for diagnostic suspicion. Genetic testing might confirm the diagnosis, with typical mutations in desmosome proteins, such as plakophilin.[2] Therapeutic approach must be individualized and sudden cardiac risk stratification should include genetic testing results, as these might indicate specific increased arrhythmic risk beyond LVEF impairment.[3]

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