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Complex Management of Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy

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Abstract : Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy (ARVD/C) is an uncommon, inheritable cardiac disorder characterized by the progressive substitution of cardiac myocytes by fibro-fatty tissues. This pathologic substitution predisposes patients to ventricular arrhythmias and right ventricular failure. The underlying genetic defect predominantly involves genes encoding for desmosome proteins, particularly plakophilin-2 (PKP2). These aberrations lead to impaired cell adhesion, heightening the susceptibility to fibrofatty scarring under conditions of mechanical stress. Primarily, ARVD/C affects the right ventricle, but it can also compromise the left ventricle, potentially leading to biventricular heart failure. Clinical presentations can vary, spanning from asymptomatic individuals to those experiencing palpitations, syncopal episodes, and, in severe instances, sudden cardiac death. The establishment of a diagnostic criterion specifically tailored for ARVD/C significantly aids in its accurate diagnosis. Nevertheless, the task of early diagnosis is complicated by the disease's frequently asymptomatic initial stages, and the overall rarity of ARVD/C cases reported globally. In some cases, as exemplified by the adult female patient in this report, the disease may advance to terminal stages, rendering therapies like Ventricular Tachycardia (VT) ablation ineffective. This case underlines the necessity for increased awareness and understanding of ARVD/C to aid in its early detection and management. Through such efforts, we aim to decrease morbidity and mortality associated with this challenging cardiac disorder.

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