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Arrhythmogenic right ventricular cardiomyopathy a diagnostic challenge in young: A case report

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ABSTRACT

Clinical presentation of Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) usually is nonspecific in young, representing a challenge diagnostic. We present an 11-year-old male patient, with a syncope event and normal ECG in the first medical assessment. Eight months later, presented a new syncope event, but now with 1st degree block that evolved to complete AV block in 24 hrs in ECG. In Tertiary Medical Care Hospital showed sinus arrhythmia with nodal rescues. The echocardiographic assessment showed dilatation and global hypokinesia in the Right Ventricle. The MR showed in apex and lateral wall late Gadolinium enhancement indicating fibrosis greater than 20%. The electrophysiologic assessment showed a low-voltage zone in the RVOT, normal stimulation between sinus and AV nodes. Was implanted ICD which identified events of ventricular tachycardia (185 bpm), giving discharges between 21 to 41 Jules, which were not perceived for the patient. Currently continues under cardiology surveillance to eventually receive a heart transplant as definitive treatment

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Introduction

The Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) is a dysplasia characterized by myocardial tissue substitution by fibro-adipose tissue associated to mutation in chromosome 1 (1q42-q43) and 14 (14q12-q22) that encode adhesion proteins (Plakoglobin, Desmoplakin, Placofinina-2 and Desmoglein-2). Is hypothesized that decoupling desmosomes in the intercalated discs of cardiomyocytes cause alteration in cell-cell adhesion or by the chromosomes mutation that encode RYR2 receptor and transforming growth factor-B3 that cause disorders in the fibrosis production stimulated by cytokines (1, 2).

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In young patients, paroxysmal ventricular tachycardia associated to syncope and nonspecific Electrocardiographic (ECG) findings are usual but the ARVC frequently is unnoticed although always must be considerate the criteria propose by Europe of Cardiology Society and Scientific Council on Cardiomyopathies (3-6). We present a case with nonspecific clinical and electrocardiographic manifestations that suggested initially a confusing diagnosis.

Case presentation

An 11-year-old male patient with an initial diagnosis challenge when presenting an isolated syncope event and apparently normal ECG in the first medical contact. Eight months later, presented a new syncope event but now with 1st degree block that evolved to complete AV block in 24 hrs in ECG. He´s sent to Tertiary Medical Care Hospital, where present sinus arrhythmia with nodal rescues and RBBB and posterior fascicle in ECG. The echocardiographic assessment showed the following findings: Dilatation and global hypokinesia of RV, interventricular septal dyskinesia, RV/LV = 0.5, E/E´= 0.87 and RVEF of 42%. The Magnetic Resonance showed myocardial fibrosis greater than 20% in apex and lateral wall with late Gadolinium enhancement (Figure 1).

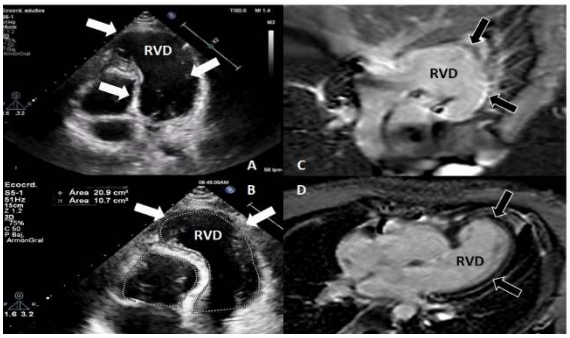

Figure 1: Echocardiographic images in 4 chambers projection (A, B) and Magnetic Resonance (B, C) that show a Right Ventricle Dliated (RVD); absence of myocardial tissue in apex, thinning and fibrosis in lateral wall (Arrows)

The electrophysiologic assessment showed a low-voltage zone in the Right Ventricular Outlet Tract, normal function in sinus node (baseline CL 626 ms at 95 bpm, SRNT 28 ms) and A-V node (AWP 350 and RWP 420 ms), with normal curve of stimulation between these nodes. In collegiate session is diagnosticated ARVC and

is implanted a Cardioverter Defibrillator (ICD), which detected some events of ventricular tachycardia (185 bpm), giving discharges between 21 to 41 Jules by ICD, that were not perceived by the patient.

Discussion

The nonspecific symptomatology observed in congenital cardiopathies with anatomical and functional disturbance, usually go unnoticed in young patients, representing a challenge diagnostic. In these cases, like patient that we present, usually beginning a complex diagnostic protocol until show cardiovascular symptoms with severe hemodynamic effects as arrhythmia and syncope. For that, we consider that in any young patients with a syncope event, always must be started a protocol assessment in Tertiary Medical Care with capacity to performance complex studies

Is true, that in young people, the variation in cardiac rhythm usually obey to physiologic or metabolic disturbance, however, always is necessary keep in mind the clinical differences of cardiopathies with anatomical and functional heart disturb with high risk of dead as ARVC and Uhl´s disease. In the Uhl´s disease, the first manifestation is a progressive heart failure but without arrhythmias and contrary in ARVC the first manifestation are arrhythmias and syncope, with heart failure in ending stages (7-12).

The histopathological assessment in ARVC show myocardial tissue substitution by fibro-adipose tissue especially in lateral wall and apex, contrasting with myocardial tissue absence between endocardium and epicardium in Uhl´s disease (7). Nevertheless, this histological approach only is possible in necropsy or biopsy, that in many cases is not an accessible procedure, so that, the images studies bring anatomical and functional information to identify both pathologies. The echocardiogram images show global dilatation, severe dyskinesia and thinning myocardial wall in RV, affecting in ARVC mainly in apex and global thinning ventricular walls in Uhl´s disease. The Magnetic Resonance is very helpful to differentiate this cardiac disease, showing in ARVC the amount of fibrotic tissue through late Gadolinium enhancement and absence of

Myocardial tissue in Uhl´s disease (6, 7). In young patients with ARCV, the ICD implantation can resolve the ventricular tachycardia events, avoiding sudden death and providing a better quality of life, allowing

patient growth to Cardiac Transplantation as definite treatment. Currently, the patient's evolution has been good with the applied treatment and continue in cardiological follow up to eventually heart transplantation as definitive treatment.

Conclusion

Adolescents with nonspecific clinical and electrocardiographic findings represent a challenge diagnostic that requires a highly specialized cardiological approach to identify opportunely anatomical and functional disturbance with high potential of sudden death as the Arrhythmogenic Right Ventricular Cardiomyopathy.

Conflicts of Interest

The authors declare that they have no competing interest regarding the publication of this study.

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