Supraventricular tachy-arrhythmia revealing an arrhythmogenic right ventricle cardiomyopathy in an adolescent: Case Report

ABSTRACT:

Arrhythmogenic right ventricular cardiomyopathy is a genetic disease that affects young people. As its name indicates, it carries a risk of ventricular rhythm disturbances and sudden death, particularly during sporting efforts emphasizing the need for early diagnosis of the disease. The most frequently observed symptoms are palpitations, syncope, clinical signs of right heart failure and cardiac arrest. These clinical manifestations, as well as the progression and prognosis of the disease have been well studied and described in the literature in adult population unlike in children where it has not been well elucidated. Its diagnosis is based on the morphological characteristics of the right ventricle during cardiac ultrasound and especially on cardiac MRI. The extent of morphological abnormalities and the frequency of occurrence of rhythm disorders make it possible to stratify the disease risk. High-risk patients should benefit from implantable cardioverter defibrillator implantation. Treatment is based on moderate physical exercise, beta blockers medical therapy and an implantable cardioverter defibrillator in certain cases.

We report a case of a 15-year-old child, with a history of first-degree consanguinity and undocumented heart disease in the father who consulted for chest pain over 8 days. The clinical presentation, electrical signs, cardiac ultrasound and imaging made it possible to make the diagnosis of ARVC.

Keywords: Arrhythmogenic right ventricle cardiomyopathy, Task Force criteria, cMRI, ICD

INTRODUCTION :

“Arrhythmogenic right ventricular dysplasia (ARVD/ARVC) is a cardiomyopathy of genetic origin caused by abnormalities of the desmosomes, characterized structurally by a progressive myocardial atrophy with fibro-fatty replacement of the right ventricle myocardium and clinically by symptoms and signs of heart electrical instability leading to ventricular arrhythmias” [1]. Numerous studies have determined that young age, male sex, non-sustained ventricular tachycardia (NSVT), syncope, extent of T wave inversion, frequent premature extrasystoles, and reduced biventricular ejection fraction are poor prognostic factors [2]. Diagnostic criteria have been well established for the diagnosis of ARVD. Cardiac imaging, in particularly cardiac magnetic resonance imaging (MRI), plays an important role in the diagnosis work-out.

CASE PRESENTATION:

We report a case of a 15-year-old adolescent, from a consanguineous marriage and with a family history of unspecified heart disease in the father, complaining for several months of palpitations which caused discomfort especially during minimum effort associated with chest discomfort, malaise and sometimes vomiting. The patient was hospitalized in the cardiological intensive care unit due to the incidental

discovery of a spontaneously reduced large QRS supraventricular tachycardia associated with stage III dyspnea of the New York Heart Association scale (NYHA). The patient reports no notion of syncope or lipothemia. The physical examination reveals a Systolic Blood Pressure (SBP) of 110mmHg and a Diastolic Pressure of 75mmHg, a heart rate (HR) at 150 beats per minute (bpm), a temperature of 36.6°C, he had a good saturation percentage value in ambient air (saturation 98%), The rest of the clinical examination was unremarkable.
Lab assessment findings: confirmed a mild anemia (Hb: 10.9g/dL), Fasting blood sugar level: 0.81g/l, cytolysis with liver enzymes twice their absolute normal values whereas the renal function, blood ionogram, C-reactive protein (CRP), as well as the hemostasis and thyroid assessment were correct.
The electrocardiogram (ECG) reveals a supraventricular tachycardia at 150bpm with large QRS complexes, a completely right bundle branch block with the presence of the epsilon wave (Figure 1).
A 24-hour Holter ECG was performed showing episodes of non-sustained ventricular tachycardia (NSVT).

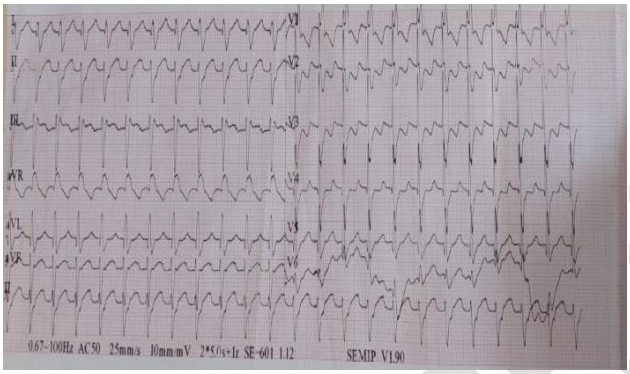

Figure 1: Supraventricular tachycardia, with the presence of a right bundle branch block and epsilon wave suggesting the diagnosis of ARVD

Transthoracic echocardiography showed a dilated, non-hypertrophied right ventricle (RV) with numerous trabeculations and an apical aneurysm with very impaired hypokinetic systolic function (S'RV at 8cm/s, right ventricular shortening fraction 28%) (Figure 2), The non-dilated and normo-kinetic wall motion left ventricle with an ejection fraction (LVEF) estimated at 50%.
Cardiac MRI reveals dilation of the right ventricle, with the presence of very significant intramyocardial fibrosis of the free wall and the tip of the RV (Figure 3, 4). The left ventricle is non-dilated, with preserved systolic function at 52%.

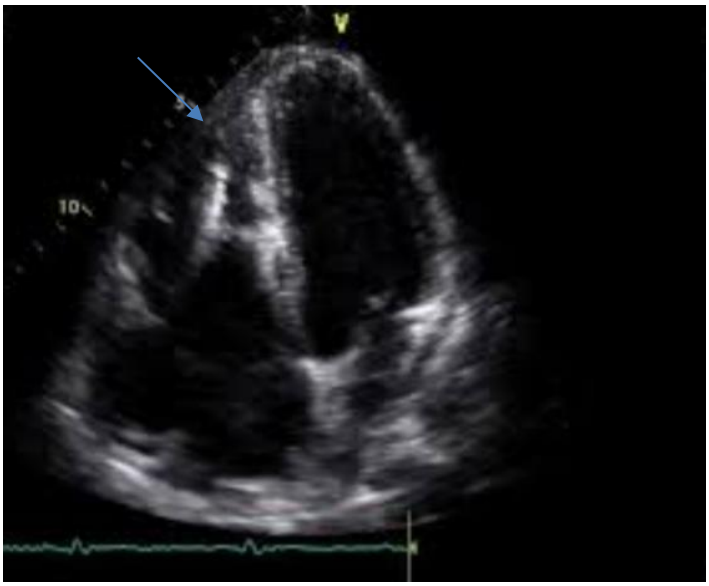

Figure 2: 4 chambers apical view; showing dilated right heart chambers with numerous trabeculations (blue arrow)

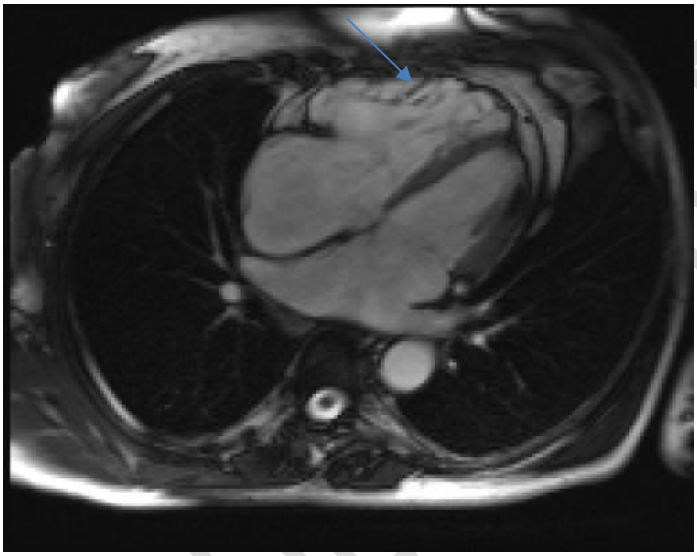

Figure 3: cardiac resonance magnetic (MRI): transverse section; showing fatty infiltration of the free wall of the right ventricle (blue arrow)

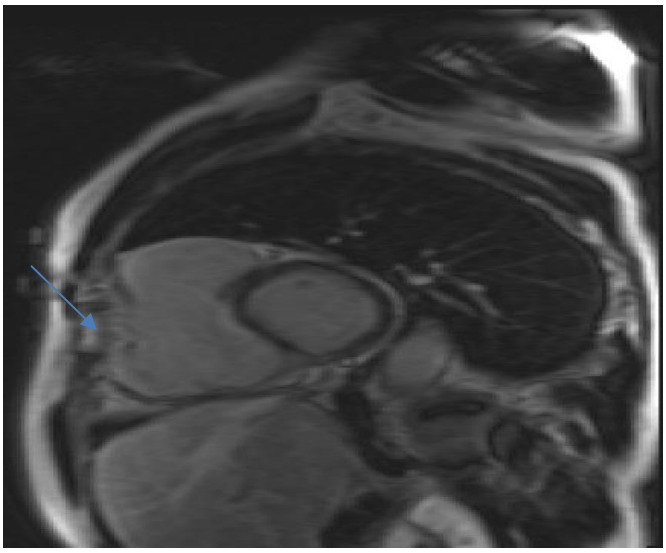

Figure 4: cardiac resonance magnetic (MRI): Sagittal sections showing the apical RV aneurysm (blue arrow)

DISCUSSION:

"ARVC/ARVD is a genetically determined heart disease, as one or more first-degree relatives also show signs of the disease in 30–50% of cases" [3].
"The vast majority of mutations observed in patients with ARVC were found in genes encoding different components of the cardiac desmosome, namely plakophilin 2 (PKP2), desmocollin 2 (DSC2), desmoglein 2 (DSG2), desmoplakin (DSP) and plakoglobin (JUP), suggesting that ARVC is a disease primarily characterized by disruption of desmosomal function. However, mutations in other genes (non-desmosomal genes) have also been reported in ARVC, including transmembrane protein 43 (TMEM43), desmin (DES), and titin (TTN), indicating genetic heterogeneity" [4]. "Several cases of ARVC have been found to be caused by multiple mutations in the same gene (compound heterozygosity) or by mutations in different genes (digenic inheritance), which could lead to earlier onset and greater severity of the disease" [5].

The benefit of early diagnosis of ARVC lies in the primary prevention of sudden death (SD), which is the most dramatic form of presentation. Indeed, ARVC is an important cause of sudden death which could be the first manifestation of the disease especially in young and particularly athletic subjects. ARVC has a prevalence that varies between one person per 2,000 to one person per 5,000 in the general population, with a clearly male predominance and a sex ratio of 3 men to 1 woman [6]. Patients with ARVC generally present between the second and fifth decades, something which was proven by a French series of 130 cases, which found an average age onset of the disease at 31.8 ± 14years with 77% of cases male sex [7]. Signs and symptoms frequently found are palpitations, syncope, ventricular tachycardia, heart failure and more severely sudden death. The same symptoms were described in children aged 3 to 16 years (category of our patient) in several case reports [8]. The presentation and course of the disease vary considerably between patients, which is probably explained by the genetic heterogeneity of the disease [9]. "The risk factors for sudden death are: history of ventricular rhythm disorders, syncope, young age, intensive physical exercise, left ventricle (LV) involvement and the absence of effective antiarrhythmic treatment" [10]. "The diagnosis of ARCV is based on a range of arguments; The diagnostic criteria for

ARCV were clarified in 1994 by the International Task Force of Cardiomyopathies and updated in 2010, and the 2010 Criteria are revised as the Padua criteria in 2020. A scoring system was established, with major (2 points) and minor (1 point) criteria, taking into account ECG, para-clinical, personal and family parameters. The diagnosis is considered certain in the presence of a score of 4, as probable if the score is 2 or 3 and as possible if the score is 2. However, these diagnostic criteria have limitations due to phenotypic heterogeneity of ARCV and incomplete penetrance. The ECG can show in the right precordial leads; localized conduction disturbances of the right ventricle, an epsilon wave (this is a deflection occurring after the end of the QRS complex, its prevalence is 30%), better visible on a high amplification ECG. Inversion of the right precordial T waves can be seen in extended forms. The diagnosis of ARVD is based on the association of these electrical abnormalities and morphological abnormalities of the right ventricle confirmed by invasive and non-invasive imaging methods"
[11, 12].
Echocardiography can reveal a decrease in contractility, a dilation of the right cavities, abnormalities of segmental kinetics, a dilation of the RV outflow chamber, and a thinning of the right ventricular wall with sometimes associated LV involvement. Magnetic Resonance Imaging (MRI) has become the reference examination in the diagnosis of ARCV, its role in the diagnosis was better described by Etoom et al. [13] who highlighted that this diagnostic tool is more useful than echocardiography in the context of the revised task force criteria.

Therapeutic management aims to: reduce patient mortality, limit progression of the disease, improve quality of life, and prevent progression to heart failure. It is based on changing your lifestyle by avoiding any intense effort; Competitive sport have shown that the increase the risk of SD five times in adolescents and young adults with ARCV [14,15]. Pharmacological treatment includes antiarrhythmic agents, beta-blockers and treatment for heart failure.

Antiarrhythmic drugs reduce the frequency and duration of arrhythmias, but no current data suggest the risk of sudden death reduction. Amiodarone, alone or in combination with beta-blockers, is the most effective drug for preventing symptomatic ventricular arrhythmias [16].

The use of beta-blockers is based on their effectiveness in preventing ventricular arrhythmias and their effectiveness in heart failure. Catheter ablation is a therapeutic option for patients with ARVC and incessant VT. The implantation of an implantable cardioverter defibrillator (ICD) in patients with ARCV should be considered as to its effectiveness and reassuring safety, especially in patients with high-risk features or a first-degree relative [17, 18].

CONCLUSION:

ARVC, a genetic disease, can manifest itself as ventricular rhythm disorders causing sudden death. The diagnosis is based on multiple criteria. Knowledge of the risk factors for the occurrence of major arrhythmic events which determine the prognosis of the disease, makes it possible to propose the best therapeutic strategy and to establish the indication for implantation of ICDs.

Consent

As per international standards, parental written consent has been collected and preserved by the author(s).

Ethical Approval:
As per international standards or university standards written ethical approval has been collected and preserved by the author(s).

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