

Case Report

CASE REPORT OF A 17 - YEAR OLD GIRL WITH ATYPICAL CLINICAL PRESENTATION OF - ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY

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Abstract. We report a case of a 17-year-old girl diagnosed with arrhythmogenic right ventricular cardiomyopathy (ARVC), who has been wrongly considered as having idiopathic ventricular extrasystoles for 13 years. The only noteworthy clinical finding until the final diagnosis was made, were complex ventricular arrhythmias (VA) during exercise as well as nonspecific repolarisations changes in inferolateral leads. We would like to increase paediatricians alertness to children presenting with so called "idiopathic" exercise induced VA that at time could turn out to be markers of arrhythmogenic condition.

Key words: arrhythmogenic right ventricular cardiomyopathy, ventricular arrhythmias

Case report

Here we report a case of a 17-year-old girl diagnosed with ARVC, who has been wrongly considered as having idiopathic ventricular extrasystoles for 13 years.

The patient was initially presented at age 4, with abdominal discomfort and feeling of irregular heartbeat.

At that time her ECG revealed ventricular ectopic beats (VEBs) of left bundle branch morphology and inferior axis; 24-hour Holter electrocardiogram revealed merely isolated VEBs but also alternative and repetitive forms with total number amounting to 1500/ 24 hours (Figs 1,2).

Other clinical examination (echocardiography, hematology, serum *biochemistry*) was unremarkable.

She was considered as having idiopathic ventricular arrhythmia and an *annual reevaluation follow-up* by a cardiologist was defined.

At age 9, exercise stress test was performed and VEBs at maximal HR of 192/min.

Until 2014 (age 15) the results of cardiovascular examination were almost unchangable. In the same year one ventricular couplet during maximal exercise stress test was noted (Fig 3) and a thorough echocardiographic examination *showed* a dilated right ventricle outflow and

inflow tracts together with enhanced apical RV trabecularisation (Figs 4 a,b,c). The patient finally underwent a cardiac MRI which definitely confirmed clinical suspicion of arrhythmogenic right ventricular cardiomyopathy (Fig. 5).

Arrhythmogenic right ventricular cardiomyopathy (ARVC) is an inherited heart muscle disease characterized by arrhythmias of right ventricle (RV) origin, due to transmural fatty or fibro-fatty replacement of atrophic myocardium [1].

In general, the clinical expression of ARVC is normally postponed until youth and adulthood and the diagnosis is seldom confirmed under the age of 10. Although there is no definite diagnostic standard, the best approach to reaching the diagnosis of ARVC is combining different diagnostic tests and clinical presentations.

The diagnosis of ARVC in children is especially difficult to establish due to the broad spectrum of phenotypic variations as well as nonspecific and inconclusive clinical findings in this age group. Inverted T waves in the right precordial leads (V1–V3) on ECG are normal findings until the age 12 and MRI is of low-yield as the anatomical, histological, and functional changes are frequently subtle or not present in the early phase of the disease [2–4]. On the other hand, right ventricular assessment by transthoracic echocardiography is by far less accurate than cardiac magnetic resonance. Hence, many ARVC cases in childhood are also diagnosed at autopsy after the occurrence of SCD [5,6].

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Received February 17th, 2016 / Accepted February 23rd, 2016

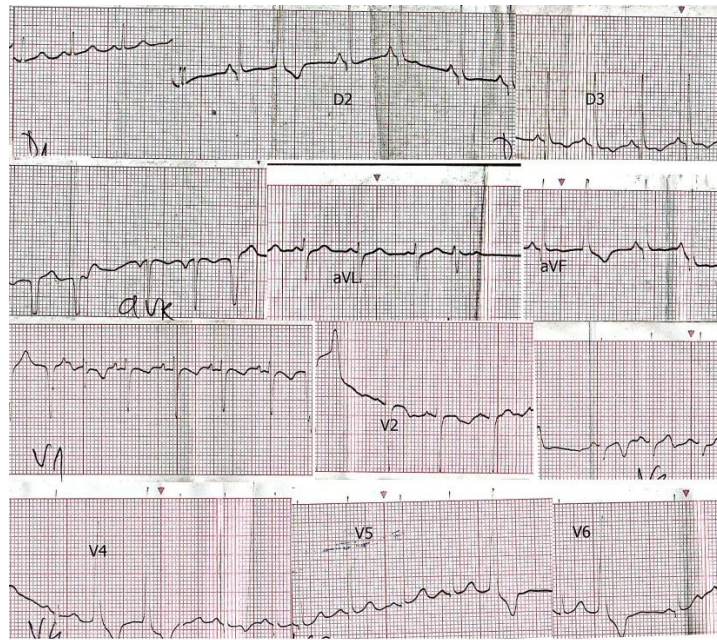


Fig. 1 Ventricular ectopic beats of left bundle branch morphology and inferior axis.

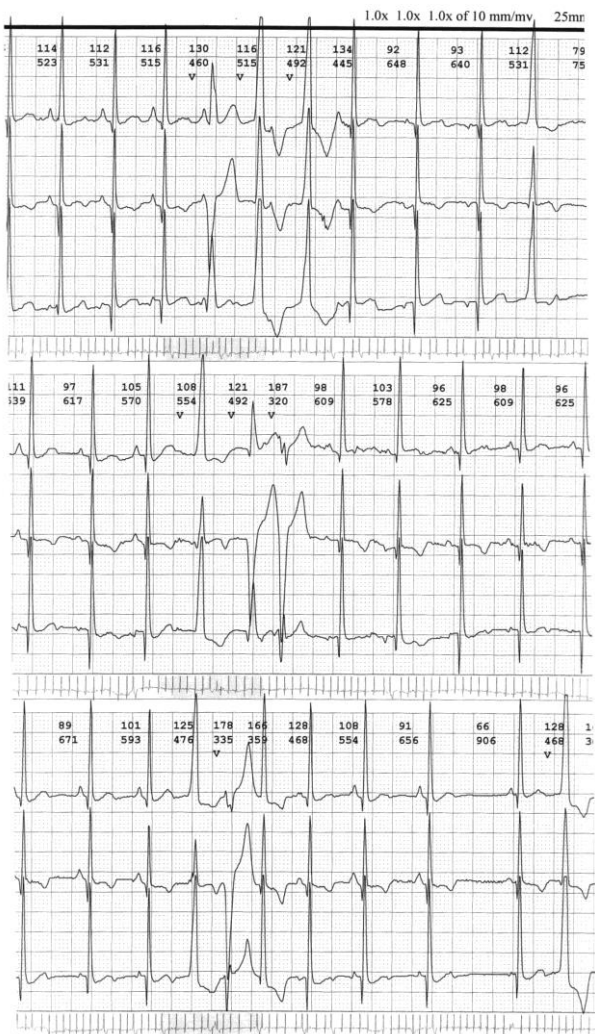


Fig. 2 Repetitive VEBs on 24 ECG Holter

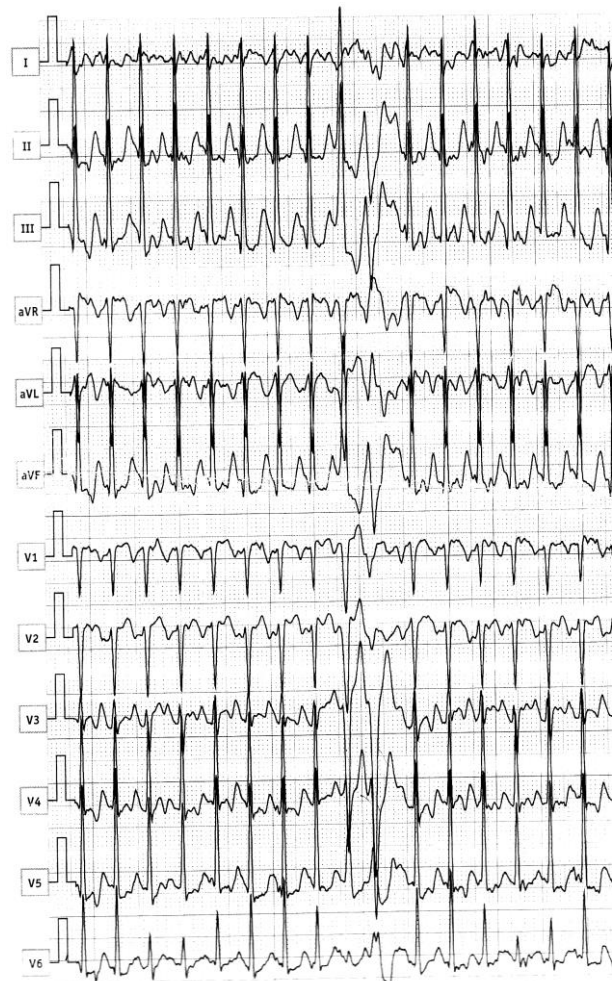


Fig. 3 Ventricular couplet during maximal exercise stress

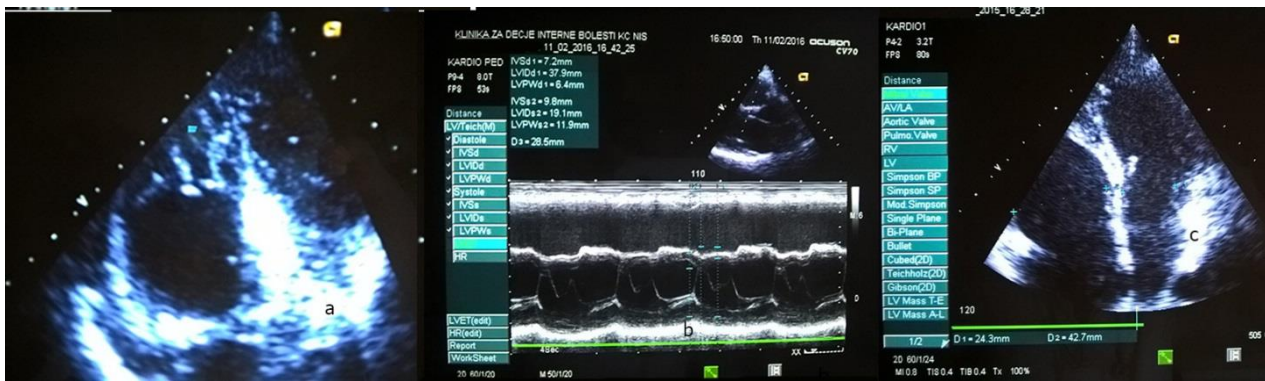


Fig. 4 a – moderator band hyper-reflectivity; b – dilation of right ventricle outflow tract (PLAX/BSA > 19 mm²); c – dilation of right ventricle inflow tract.

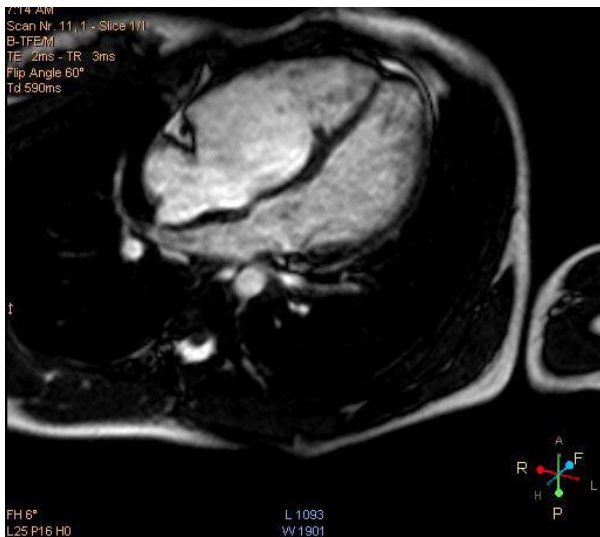


Fig. 5 Aneurismal outpouchings of RV on MRI

Recent research suggests that electrical abnormalities in the form of complex VEBs or VT precede structural changes in ARVC. Of them ventricular tachycardia with LBBB morphology and superior axis is the most characteristic presentation in children.

Among other pathologic findings, echocardiographic abnormalities (dilated and hypokinetic, akinetic or dyskinetic right ventricle) even in asymptomatic patients, should arouse suspicion of this cardiomyopathy. Right ventricle endomyocardial biopsy, if positive, is a gold standard for diagnosis, but often yields a false-negative result (sensitivity is approximately 67%) [7].

Of note is that ARVC in our patient had begun insidiously. Except for the presence of complex VAs (left bundle branch block and inferior axis disappearing at submaximal HR during stress test) at the time of disease presentation, all the other clinical and cardiovascular examinations were normal. The arrhythmias were considered idiopathic and the patient was free of symptoms over months to years although she had been an active football player from age 9 to age 15.

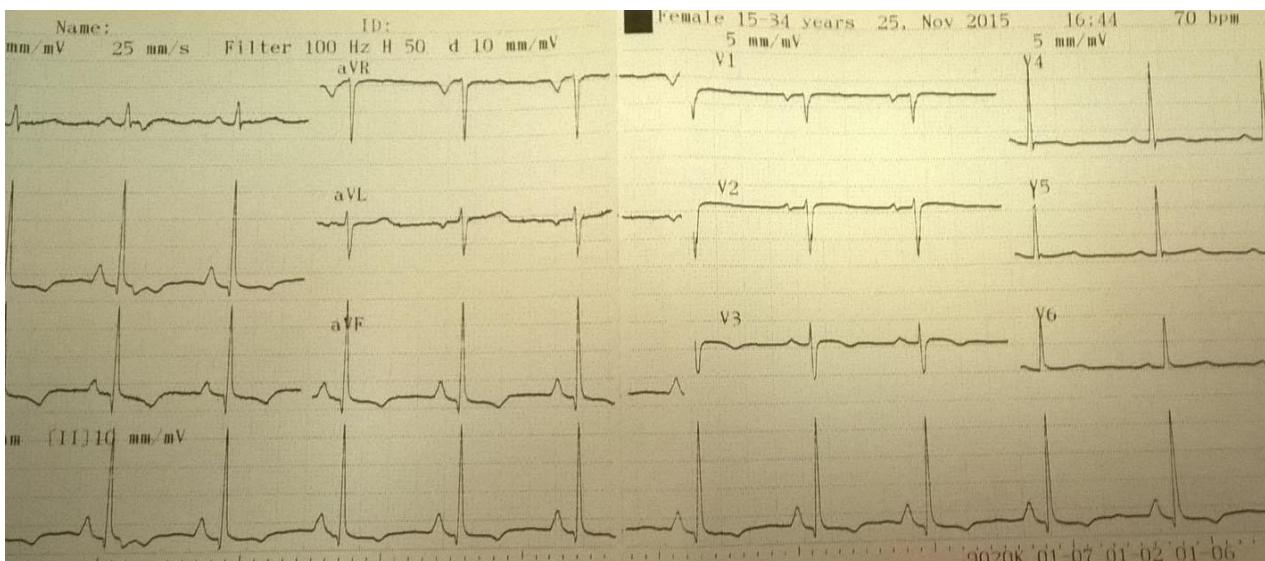


Fig. 6 Nonspecific repolarisation changes in inferolateral leads

Distinctively, her 12 leads ECG was not suggestive of ARVC at any time of the disease course and only nonspecific repolarisation abnormalities in left precordial leads were noticed at age 12 (Fig 6). The more suggestive clinical characteristics in the form of pathologic major echocardiographic criteria that facilitate the recognition of ARVC were not discernible until the patient reached 15 years (Fig 4 a,b,c).

Of note is that in contrast to the apparently non-life-threatening implication of idiopathic ventricular arrhythmias (IVAs) at rest, IVAs elicited during exercise, even in apparently normal individuals, appear to imply risk over time [8]. To date, a number of publications have confirmed that IVAs might be the initial and unique manifestation of clinically silent arrhythmogenic conditions such as myocarditis [9, 10]. On the other hand it is well known that a large majority of patients with ARVC have histological evidence suggestive of inflammation [11].

The only noteworthy clinical finding in our patient, until the occurrence of typical echocardiographic

changes, were complex VAs during exercise as well as nonspecific repolarisation changes in inferolateral leads on ECG (Fig 6).

However, their clinical significance was not questioned as the patient was asymptomatic during routine follow-up, despite actively plying football for a decade. Although of not yet defined clinical significance, exercise induced VA is not a common finding in children with benign IVAs etiology. In our opinion, the early appearance of VA (VEBs) is unlikely due to right ventricle structural changes and might be explained more simply by assuming that their origin is a consequence of an inflammatory process which modulated the clinical progression of ARVC in our patient.

With this case report, we would like to increase paediatricians' alertness to children presenting with so called "idiopathic" exercise induced VA, that are probably the earliest biomarkers of more severe arrhythmogenic condition. Prolonged reevaluation follow-up of children with IVAs by pediatric cardiologist should be recommended in similar cases.

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