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Case Report

Sudden Cardiac Death due to Arrhythmogenic Right Ventricular Dysplasia

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ABSTRACT

We report a case of sudden cardiac death of young man in which right ventricular dysplasia/cardiomyopathy was revealed by post mortem examination. Arrhythmogenic Right Ventricular Dysplasia/cardiomyopathy is of unknown etiology seen in upto 1-2.5% of individuals who experience sudden cardiac death, mainly athletes. The histomorphological explanation of such death was a lipomatous or fibrolipomatous replacement and infiltration of the myocardium of the right ventricle. It was suggested that death is due to electrical instability of the right ventricular myocardium leading to development of tachyarrythmias which ended in ventricular fibrillation and consequently death. There were no congenital anomalies nor signs of myocardial degeneration, necrosis or inflammatory infiltration noted.

Keywords: Arrhythmogenic right ventricular dysplasia, Cardiomyopathy, Sudden cardiac death.

INTRODUCTION

Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy (ARVD/C) is a distinct clinical and morphologic entity which was first reported in 1982 by Marcus et al in the medical literature. [(1)] ARVD/C is an important cause of unexpected sudden death among young people especially in athletes. Although the true incidence and prevalence of ARVD/C are unknown they are estimated at 1-5,000 with significant familiar occurrence.[(2)] The disease is an inherited non ischemic cardiomyopathy characterized pathologically by fibrofatty replacement affecting primarily the right ventricular myocardium. [(1)] This article reviews recent advances with respect to genetics, pathophysiology and manifestation of disease, which was an incidentally encountered autopsy finding.

CASE HISTORY

A 32 year old male, presented with acute chest pain and was taken to the hospital where he was declared dead on arrival. The deceased was known to be suffering from asthma and was on treatment. Post mortem was carried out and the organs retrieved were sent in formalin for histopathological examination. Heart weighed 262g.Externally, epicardial fat was increased. On opening the heart along the flow of blood- right atrium, right ventricle, right atrioventricular septum, right chordae tendinae and papillary muscle showed yellowish colouration. Right ventricular wall

measured 2mm and appeared thinned out (Fig1). Epicardial fat thickness measured 5mm.Right coronary artery appeared unremarkable. Left atrium and left ventricle appeared unremarkable. Left ventricular wall thickness measured 17mm .Tricuspid valve circumference was 13cms and bicuspid valve - 12cms.Aorta showed multiple pinpoint sized yellowish fatty streaks. Left coronary vessels were patent and thickened.

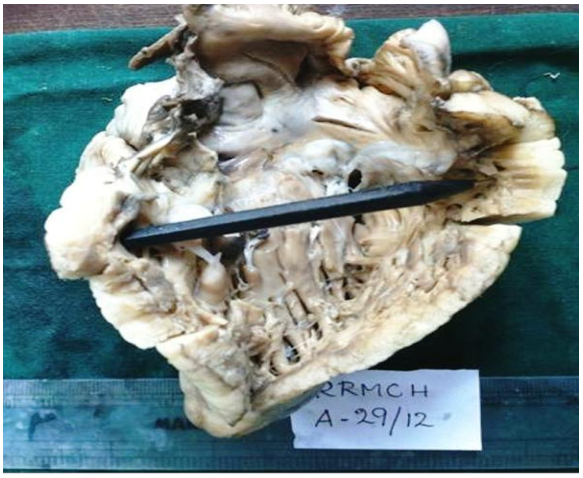

Fig1:Thinned out right ventricle with increased fat deposition.

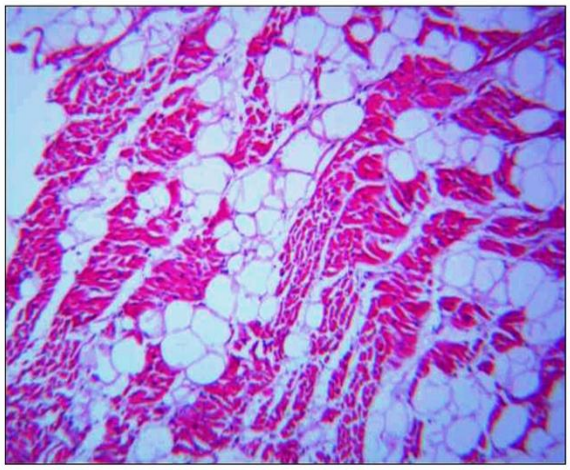
Fig 2: Lipomatous infiltration into the right ventricular cardiomyocytes (H&E X10)

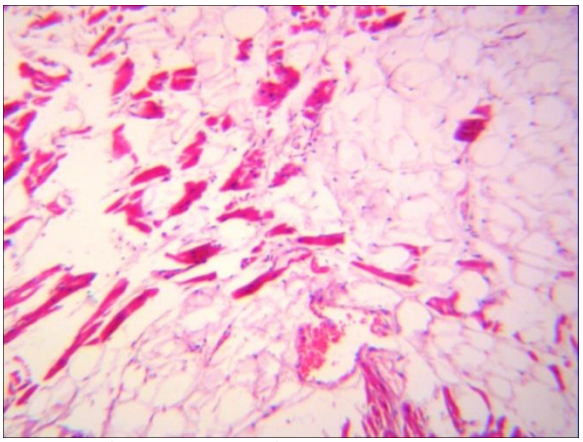
Fig 3: Multifocal lipomatous infiltration into the right papillary muscle (H&E X10).

Microscopically, right ventricular wall, right chordae tendinae, right papillary muscle, right atrium, left atrium and right atrioventricular septum showed areas of fat infiltration in the perivascular region as well as between myocardial fibres (Fig2 &3)The left ventricular wall , left chordae tendinae, left papillary muscle showed no evidence of fatty infiltration. Right and left atrial appendages showed evidence of fatty infiltration. Aorta, left coronary artery and right coronary artery showed evidence of atheromatous plaque.

Right Lung tissue was congested, spongy and weighed 300 gms. Microscpically, right lung showed evidence of pulmonary oedema and congestion. There were no other significant findings in the left lung, pancreas, adrenals, spleen, cerebrum grossly and microscopically. The diagnosis of Arrythmogenic Right Ventricular Dysplasia/ Cardiomyopathy was made from the above conglomeration of findings.

DISCUSSION

Sudden death is defined as death of an apparently healthy person who is not known to be suffering from any disease, injury or poisoning is found dead or dies within one hour after the onset of terminal

illness. Most cases of sudden death in the young is due to infection ,epilepsy, intracranial hemorrhage and asthma .Unrecognized structural or functional heart disease such as hypertrophic cardiomyopathy, dilated cardiomyopathy, myocarditis, acute myocardial infarction, congenital anomalies, arrythmogenic right ventricular dysplasia/ cardiomyopathy and premature coronary artery disease are the causes of Sudden Cardiac Death in young. [(3)]

ARVD/C accounts for 1-2.5% of sudden cardiac deaths especially among athletes. It is essential to be aware of these rare anatomical conditions. ARVD/C is a heritable condition typically with an autosomal dominant pattern of inheritance and variable penetrance. Different genetic variants of ARVD/C have been mapped. The disease loci are mapped to ARVD1, ARVD2, ARVD3 and ARVD4 in the chromosome 14q, 1q, 2q, 23q, 24q. [(4)] According to dystrophic theory, the progressive loss of myocardium is considered secondary to myocyte death due to genetic defects. More recently, cell adhesions protein like, desmoplakin and plakophilin 2 gene mutations have been found in dominant forms of ARVD/C. Impaired functioning of the cell adhesion complex under conditions of mechanical stress is thought to cause myocyte detachment and death. [(5, 6)]

An inflammatory theory has been advanced with infection and toxic or immune mechanisms have been postulated. Enterovirus, Coxsakievirus B3, and different cardiotropic viruses such as adenovirus, cytomegalovirus, hepatitis C virus, and parvovirus B19 have been detected in sporadic ARVD/C by researchers. Hence fibrofatty infiltration of ARVD/C is viewed as a healing phenomenon in the setting of chronic myocarditis caused by these organisms. [(5)]

ARVD/C is a disease of the myocardium associated with electrical dysfunction. The peculiar pathological feature consists of progressive loss of the right ventricular myocardium with fibrofatty replacement. In nearly half of the cases, the left ventricle is also involved. The thinning of the right ventricular free wall due to myocardial atrophy with fibrofatty replacement, accounts for the occurrence of aneurysms, typically located in the inflow, the apex and the outflow tract (so called triangle of dysplasia) of the right ventricle. This progressive replacement is thought to be responsible for the presence of late potentials detected with signal averaged ECG. Aneurysms of the right ventricle are a pathognomonic feature of the disease and are easily detectable at cardiac imaging. The observation of fibrofatty replacement at right ventricular endomyocardial biopsy may help in achieving an in-vivo diagnosis. [(2, 5)] The diagnosis is established based on the criteria set by the Task Force of the Working Group of Myocardial and Pericardial Disease of the European Society of Cardiology and of the Scientific Council on Cardiomyopathies of the International Society and Federation of Cardiology in which 2 Major criteria or 1 major plus 2 minor criteria or 4 minor criteria are considered diagnostic. (Table1). [(2, 6)]

The clinical presentation of ARVD/C varies from silent forms with an exercise related episode of syncope or sudden death as first manifestation to biventricular cardiac failure that requires cardiac transplantation. Common symptoms include palpitation during physical activity, fatigue, dizziness, atypical chest pain and one or more episodes of syncope. [(4)] Progression and extension of the disease could also provoke right and /left ventricular failure which could lead to haemodynamic arrhythmic death. [(2, 4- 6)]

TABLE 1: DIAGNOSIS OF ARVD/C

	Major Criteria	Minor criteria
1.Structural or functional abnormalities	1.Severe dilatation and reduction of RVEF* 2.Localized right ventricle aneurysm 3.Severe segmental dilatation of the right ventricle	1.Mild global right ventricular dilatation and/or EF reduction 2. Mild segmental dilatation of right ventricle 3. Regional right ventricular Hypokinesia.
2.Tissue characterization	Infiltration of right ventricle by fat with the presence of surviving strands of cardiomyocytes	
3.ECG** depolarization/conduction abnormalities	1.Localized QRS complex, duration > 110ms, in V1, V2 or V3. 2.Epsilon waves in V1, V2 or V3	Late potentials in SAECG[#]
4.ECG repolarization abnormalities		Inverted T waves in right precordial leads.
5.Arrhythmias		1. Ventricular tachycardia of LBBB[$] morphology 2. Frequent ventricular extrasystoles(1000/24hr on holter)
6.Family history	Family history of ARVD confirmed by biopsy or autopsy.	1.Family history of premature sudden death(<35 years) due to suspected ARVD/C 2. Family history of clinical diagnosis based on present criteria.

*Right ventricular ejection fraction; **Electrocardiogram; # Signal Average Electrocardiogram; $ Left Bundle Branch Block

Sudden death unfortunately may be the first manifestation of the disease, when ventricular tachycardia degenerates into ventricular fibrillation. [(5)] It occurs with no previous critical signs, and post mortem examination is mandatory to clarify the cause of death. Even in autopsies, this disease may often be overlooked because the pathologist is unaware of its existence. The right ventricle should be extensively sampled for histopathologic analysis in all cases of sudden death, especially those associated with strenuous exercise and young age.

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