INTRODUCTION

Arrhythmogenic right ventricular dysplasia (ARVD), or arrhythmogenic right ventricular cardiomyopathy (ARVC), is an uncommon form of inheritable disease of the myocardium. It is most commonly associated with right ventricular failure and various rhythm disturbances, particularly ventricular tachycardia or fibrillation. Left-sided involvement with left-sided heart failure may also occur. It is a recognized cause of sudden cardiac death in the young athletes.

In the present case series, we reported 2 cases of arrhythmogenic right ventricular dysplasia (ARVD). One of whom died due to electrocution while other was sudden death. Both were young males. Both males were manual labourer and used to do hard labour. Both were chronic alcoholic. The diagnosis of ARVD was made during autopsy and histopathological examination of the heart. There was fat infiltration in the myocardium till the endocardium of right ventricle, left ventricle and inter ventricular septum.

Arrhythmogenic right ventricular cardiomyopathy (ACM), Sudden cardiac death, Young athletes, Inheritable disease of the myocardium, fat infiltration in the myocardium.

ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA/CARDIOMYOPATHY – A CASE SERIES

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ABSTRACT

Arrhythmogenic right ventricular dysplasia (ARVD), also known as arrhythmogenic right ventricular cardiomyopathy (ARVC), or arrhythmogenic cardiomyopathy (ACM) is an uncommon form of inheritable disease of the myocardium. In this condition the right ventricular wall is severely thinned due to loss of myocytes, with extensive fatty infiltration and interstitial fibrosis. It is most commonly associated with right ventricular failure and various rhythm disturbances, particularly ventricular tachycardia or fibrillation. Left-sided involvement with left-sided heart failure may also occur. It is a recognized cause of sudden cardiac death in the young athletes.

In the present case series, we reported 2 cases of arrhythmogenic right ventricular dysplasia (ARVD). One of whom died due to electrocution while other was sudden death. Both were young males. Both males were manual labourer and used to do hard labour. Both were chronic alcoholic. The diagnosis of ARVD was made during autopsy and histopathological examination of the heart. There was fat infiltration in the myocardium till the endocardium of right ventricle, left ventricle and inter ventricular septum.

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INTRODUCTION

Arrhythmogenic right ventricular dysplasia (ARVD), or arrhythmogenic right ventricular cardiomyopathy (ARVC), is an uncommon form of inheritable disease of the myocardium. It is most commonly associated with right ventricular failure and various rhythm disturbances, particularly ventricular tachycardia or fibrillation. Left-sided involvement with left-sided heart failure may also occur. It is a recognized cause of sudden cardiac death in the young athletes.

Morphologically, the right ventricular wall is severely thinned due to loss of myocytes, with extensive fatty infiltration and interstitial fibrosis. Although myocardial inflammation may be present, ARVD/C is not considered an inflammatory cardiomyopathy. Classical ARVD/C has autosomal dominant inheritance with a variable penetrance. The disease has been attributed to defective cell adhesion proteins in the desmosomes that link adjacent cardiac myocyte.

Naxos syndrome is a disorder characterized by arrhythmogenic right ventricular cardiomyopathy and hyperkeratosis of plantar palmar skin surfaces specifically associated with mutations in the gene encoding the desmosome-associated protein plakoglobin. It has autosomal recessive inheritance.1,2 ARVD was first described in 1977 by Fontaine et al during a surgery to map and treat ventricular tachycardia at the Hospital de La Salpetriere.3 It was included in the new classification of cardiomyopathies in 1996.4

In the general population, the prevalence of ARVC/D is 1 per 2500 to 1 per 5000. The prevalence is higher in Italy (Padua, Venice) and Greece (Island of Naxos). This disorder accounts for 5% to 10% of sudden unexplained death in individuals less

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than 65 years of age. It occurs in young adults and has a male
to female ratio of 2.7 to 1.5

Those affected by Arrhythmogenic right ventricular dysplasia
(ARVD), or arrhythmogenic right ventricular cardiomyopathy
(ARVC), or arrhythmogenic cardiomyopathy (ACM) may not
have any symptoms at all despite having significant
abnormalities in the structure of their hearts.6 If symptoms do
occur, the initial presentation is often due to abnormal heart
rhythms (arrhythmias) which in arrhythmogenic
cardiomyopathy may take the form of palpitations, or
blackouts.7 Sudden death may be the first presentation of ACM
without any preceding symptoms.8 These symptoms often
occur during adolescence and early adulthood, but signs of
ACM may rarely be seen in infants.

Case series

Case 1

A 26 years old male who was a laborer by occupation suddenly
had palpitations and collapsed at work place. He was taken to a
hospital where he was declared brought dead. He was an
occasional drinker and used to drink alcohol since 5-10 years,
and used to do hard labour. There was no family history of any
heart disease. Autopsy was conducted at the mortuary of
Victoria hospital, Bangalore Medical College and Research
Institute, Bangalore. On external examination the dead body
measured 160 cm in length, moderately built and moderately
nourished. Rigor mortis was present all over the body. Post
mortem staining was seen over the back of the body. There
were no external injuries on the body. On internal
examination, Heart weighed 250 grams. Right ventricle wall
thickness measured 0.5 cm. Left ventricle wall thickness measured 1.5
cm. Interventricular septum thickness measured 1.5 cm.
Yellowish fatty infiltration seen in the myocardium and
endocardium of right ventricle, left ventricle and interventricular septum at places. Coronaries were patent. Lungs showed congestion and edema. Liver showed yellowish shiny areas at places suggestive of fatty changes. Other internal organs were congested. On histopathological examination of
heart, fat infiltration seen in the myocardium till the endocardium of right ventricle, left ventricle and interventricular septum at places suggestive of Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy.

Case 2

A 28 years old male who was a manual laborer by occupation
died due to electrocution. He was chronic alcoholic and
smoker, and used to do hard labour. There was no family
history of any heart disease. Autopsy was conducted at the
mortuary of Victoria hospital, Bangalore Medical College and
Research Institute, Bangalore. On external examination the
dead body measured 155 cm in length, moderately built and
moderately nourished. Rigor mortis was present all over the
body. Post mortem staining was seen over the back of the body.
There were no external injuries on the body. On internal
examination, Heart weighed 250 grams. Petechial haemorrhages present over surface of heart at places. Right
ventricle wall thickness measured 0.5 cm. Left ventricle wall
thickness measured 1.8 cm. Interventricular septum thickness measured 2.0 cm. Yellowish fatty infiltration seen in the
myocardium and endocardium of right ventricle, left ventricle and interventricular septum at places. Grossly coronaries were patent. Ascending aorta showed fatty streaks at places. Lungs showed congestion and edema. Liver showed yellowish shiny areas at places suggestive of fatty changes. Other internal organs were congested. On histopathological examination of
heart, fat infiltration seen in the myocardium till the endocardium of right ventricle, left ventricle and interventricular septum at places suggestive of Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy. Left coronary artery showed atherosclerosis without any luminal narrowing. Other coronaries were normal and patent.

DISCUSSION

The causes of sudden cardiac death differ greatly among
various age groups. In individuals > 40 years old,
atherosclerotic coronary heart disease is the most common
cause. Between 1 to 40 years of age, the causes of sudden
cardiac death are commonly hypertrophic cardiomyopathy,
myocarditis, congenital heart disease, arrhythmogenic right
ventricular dysplasia/ cardiomyopathy etc.8

The sudden death in apparently healthy young individuals is
always a devastating and shocking event. The incidence of
sudden deaths in young due to various cardiac pathologies has
wide variations in different parts of the world. Now because of
change in lifestyle and habits, many cardiac pathologies are
noticed in comparatively younger age group even in a
developing country like India and some of these cardiac

Fig 1 The photomicrograph showing fat infiltration in the myocardium till the endocardium suggestive of Arrhythmogenic Right Ventricular Dysplasia (H & E, 10X).

Fig 2 Another photomicrograph showing fat infiltration in the myocardium till the endocardium suggestive of Arrhythmogenic Right Ventricular Dysplasia (H & E, 10X).
pathologies like arrhythmogenic right ventricular dysplasia/ cardiomyopathy are inheritable.

In a study by Eckart RE et al, in America, over a period of 10 years, in 902 cases of adjudicated unanticipated sudden cardiac death, they found that out of total 298 cases of <35 years of age arrhythmogenic right ventricular dysplasia was seen in 4 (1.3%) cases while out of 604 cases of age ≥35 years ARVD was seen in 6 (1%) cases.

Mu J et al, in a retrospective study in China between January 1986 and September 2017, studied 45 Chinese ARVC autopsy case reports from Chinese literature databases and 2 case reports of sudden death due to ARVC, observed that there were 27 males and 20 females, and the mean age at death was 35 years. Sudden cardiac death was the first manifestation observed in most patients, with no previous family and medical history. Exercise, acute stress, increased cardiac workload, and ethanol are frequently involved. Microscopic abnormalities included replacement of myocardium by adipose infiltration in 68.09% cases and fibroadipose in 31.91% cases; 80.85% cases were restricted to the right ventricle (RV), whereas biventricular subtype was seen in the remaining 19.15% cases.

Pawel BR et al, reported two cases of sudden death due to arrhythmogenic right ventricular dysplasia (ARVD) occurring in the pediatric age group. One of the subjects, at the age of 7 years, is believed to be the youngest child in whom ARVD has been diagnosed at autopsy.

Schionning JD et al, in 1997, reported five cases of ARVD identified by autopsy. In three of the cases, sudden death occurred in the young (16-28 years old) during or shortly after exercise.

In the present case series, we reported 2 cases of arrhythmogenic right ventricular dysplasia (ARVD). One of whom died due to electrocution while other was sudden death. Both were young males. Both males were manual labourer and used to do hard labour. Both were chronic alcoholic. Microscopically, there was fat infiltration in the myocardium till the endocardium of right ventricle, left ventricle and inter ventricular septum. One case was also found to have associated atherosclerotic coronary artery disease.

CONCLUSION

In this case series arrhythmogenic right ventricular dysplasia/cardiomyopathy was found during autopsy on histopathological examination of heart in 2 young individuals who were manual labourer. One of whom died suddenly, while other died due to electrocution. Finding cases of arrhythmogenic right ventricular dysplasia (ARVD), in a country like India is a matter of concern. This case series emphasizes the requirement of essential investigations to diagnose arrhythmogenic right ventricular dysplasia (ARVD) early especially in young athletes and labourer, and preventive measures to prevent sudden cardiac death in young individuals and their family members due to inheritable cardiac diseases like this in future. This case series also highlights the role of meticulous autopsy and histopathological examination to detect this condition.

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