CASE REPORT

Electrical Storm or Naxos Syndrome in an Adult Causing Recurrent Syncope
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ABSTRACT
Among the rare and well-known causes of sudden cardiac death by malignant arrhythmias is a condition called arrhythmogenic right ventricular cardiomyopathy. It commonly presents with right ventricular dilatation, dysfunction and ventricular tachycardia of left bundle branch morphology due to fibro-fatty infiltration of right ventricle in second to fifth decade of life, making it an unrecognized and important cause of sudden cardiac death. Two rare variants of arrhythmogenic right ventricular cardiomyopathy are Carvajal syndrome and Naxos syndrome. Both variants have systemic manifestations. Being a rare variant of arrhythmogenic right ventricular dysplasia, Naxos syndrome was initially described in the families of the Greek island of Naxos. It is a recessive disorder with cardio-cutaneous manifestations characterized by arrhythmogenic right ventricular cardiomyopathy, palmoplantar keratoderma and wooly hair. We report a rare case of Naxos syndrome in an adult patient presented with recurrent episodes of palpitation and syncope.


INTRODUCTION
Arrhythmogenic right ventricular cardiomyopathy is one of the causes of sudden cardiac death in young adults, caused by malignant arrhythmias and a reported incidence of sudden cardiac death per year 1 - 2%.1-3 Positive family history is present in 30 - 50% of cases and the disease usually follows an autosomal dominant pattern with variable penetrance and expressivity.2 Autosomal recessive pattern is rarely reported which has two rare variants, known as Naxos and Carvajal syndromes. Naxos syndrome is characterized by palmoplantar keratoderma and wooly hair. The cause of Naxos syndrome is mutations in desmosomal proteins resulting in fibro-fatty infiltration of right ventricle. Patient may present with syncope, ventricular tachycardia or sudden death.1-3

Diagnosis of arrhythmogenic right ventricle cardiomyopathy is based on new task force criteria which extensively comprises of ECG, echocardiographic, cardiac catheterization features and positive family history. As for Naxos syndrome, its diagnosis requires arrhythmogenic right ventricle cardiomyopathy with cardio-cutaneous manifestations palmoplantar keratoderma and wooly hair with the presence of normal coronary arteries.4 The genetic mutation responsible is deletion of two base pairs in plakoglobin genes, which is also included for definitive diagnosis.5 We report this case because Naxos syndrome is a rare variant of arrhythmogenic right ventricular cardiomyopathy, hardly reported in Pakistani population.

CASE REPORT
A 40-year male presented in emergency with recurrent episodes of palpitations and loss of consciousness. ECG showed ventricular tachycardia with left bundle branch pattern morphology, which was electrically cardioverted (Figure 1A). Patient was treated with anti-arrhythmic agents. All routine laboratory tests were normal. Resting ECG obtained after settlement of arrhythmia showed Epsilon wave (negative deflection between the end of QRS complex to the onset of

Figure 1: (A) ECG showing Epsilon wave. (B,C) Palmoplantar keratoderma.
T-wave) and T-wave inversions from V1-V3. He was also suffering from palmoplantar keratosis since childhood (Figures 1B and 1C). Echocardiogram was done, which showed right ventricular dilatation with impaired right ventricular systolic function and normal left ventricular systolic function. Coronary angiogram showed normal coronary arteries. Cardiac MRI showed fibro-fatty infiltration of the right ventricle. An intra-cardiac device was implanted considering the survival of sudden cardiac death. As patient was survived from sudden cardiac death and it is class I indication to implant an intra-cardiac device in such patients. Patient was advised not to participate in any kind of competitive support or aggressive activities and was put on beta-blockers for long-term management.

DISCUSSION
A recessive disorder and initially described in families originating from the Greek island of Naxos, Naxos syndrome is a rare variant of arrhythmogenic right ventricular dysplasia with cardio-cutaneous manifestations characterized by palmoplantar keratoderma and woolly hair. Affected families have also been reported in Saudi Arabia, Turkey, Israel and Greek Aegean Islands.5 Another variant of arrhythmogenic right ventricular dysplasia, more prevalent in the sub-continent, is Carvajal syndrome with predominantly left ventricular involvement and clinically more similar to dilated cardiomyopathy.6

Clinical presentation varies from palpitations or sustained ventricular tachycardia with left bundle branch morphology, syncope or even sudden cardiac death. Cardiomyopathy manifests at adolescence with 100% penetrance and almost 30% of patients become symptomatic before their thirties.4 Histologically, Naxos syndrome shows areas of extensive myocardial loss and replacement fibrosis; and at genetic level two base pair deletion in plakoglobin gene, which maps at 17q21 is described as the cause of Naxos syndrome. Defect in the gene results in defect in linking proteins plakoglobin and desmoplakin linking sites especially under stress, resulting in progressive loss of myocardium and fibro-fatty replacement.8 Almost all cases exhibit repolarization or depolarization abnormalities of right ventricle on echo-cardiography, leading to diagnosis of arrhythmogenic right ventricular dysplasia according to established criteria.4

The main goal of management is the prevention of sudden cardiac death. Implantation of an automatic cardiac defibrillator is indicated in patients who are the survivors of sudden cardiac arrest or who develop symptoms. Recurrent episodes of ventricular tachycardia can be prevented with anti-arrhythmics alone or in combination with beta-blockers. Systemic genetic screening for populations at risk of Naxos syndrome is also under consideration.7

REFERENCES