

Carvajal Syndrome-A Rare Variant of Naxos Disease in Two Saudi Siblings

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Abstract

Naxos disease is a recessively inherited condition characterised by woolly hair, palmoplantar keratosis and arrhythmogenic right ventricular cardiomyopathy (ARVC). A rare syndrome with similar clinical picture and predominantly involving left ventricle has been termed as Carvajal Syndrome. We report two cases of a Saudi family admitted in our PICU.

Keywords: Naxos disease; Carvajal syndrome; Cardiomyopathy

Introduction

Carvajal syndrome also known as 'Striate palmoplantar keratoderma with woolly hair and cardiomyopathy is a cutaneous condition inherited in an autosomal recessive pattern due to a defect in desmoplakin gene. The skin disease presents as a striate palmoplantar keratoderma particularly at sites of pressure. The patient is at risk of sudden cardiac death due to dilated cardiomyopathy associated with this entity. We report two cases of a Saudi family admitted in our PICU to highlight the importance of this syndrome.

Case 1

A 5 year old Saudi girl presented with cough, breathlessness and easy fatigueability for 3 days duration. On general examination she had fine, brittle, lustreless scalp hair; pallor; stomatitis, keratotic skin over palm, sole and lower half of both lower limbs. She had tachycardia with heart rate of 142/min; respiratory rate of 40/min; Blood pressure 80/60 mmHg, SPO₂ 72% in room air; Systemic examination revealed diastolic murmur grade 3/6 over mitral, tricuspid area; raised JVP with bilateral basal crepitations. There was tender hepatomegaly (5 cm) with liver span-10.8 cm, with positive hepatojugular reflex. Central nervous system was apparently normal. X-ray chest showed- Cardiomegaly with pulmonary plethora. Blood investigation revealed-CK-1122 u/l; CKMB-45.9 u/l; LDH-319 u/l. ABG showed hypoxemia with metabolic acidosis. ECG findings were suggestive of biventricular hypertrophy. 2D-ECHO showed-Dilated LV (51 mm), Dilated RV+RA; Mild Mitral as well as tricuspid regurgitation; Pulmonary hypertension (55 mmHg); Ejection fraction-39%. The patient was admitted in our PICU and was managed as a case of heart failure and was investigated for the association of her dermatologic manifestations with cardiomyopathy. The genetic study by DNA PCR revealed missense mutation in desmoplakin (DSP)s gene. The child stayed for five days in PICU and succumbed to death despite adequate measures for heart failure.

Case 2

The elder brother (10 yr age) of the index case was admitted one month later with similar manifestations of skin and presented as a case of heart failure. He expired within 2 days of admission in our PICU. His genetic study showed the similar result of missense mutation in DSP gene.

Discussion

Naxos disease was first described by Protonotarios et al in families originating from the Greek island of Naxos [1]. It is caused by a recessive mutation in desmoplakin, which maps to 17q21 gene, an intracellular protein that links desmosomal adhesion molecules to intermediate filaments of the cytoskeleton. Patient usually presents

with Woolly hair since birth; palmoplantar keratosis develops during first year of life and cardiomyopathy (Rt.ventricular) in adolescence [1-4]. A particular mutation that truncates the intermediate filament-binding site of desmoplakin results in a variant of Naxos disease with predominantly left ventricular involvement, early morbidity and clinical overlapping with dilated cardiomyopathy has been described by Carvajal-Huerta et al. from Ecuador as Carvajal Syndrome [5]. It is a progressive heart disease and may cause sudden death in a child with early age. Whenever a child presents with such a dermatological manifestation, the paediatric cardiologist's consultation must be done at earliest possible. The primary goal of the management is to prevent sudden cardiac death. Implantation of automatic cardioverter defibrillator, antiarrhythmic drugs and management of heart failure are the recommended treatment modalities [6]. The population at risk should be genetically screened.

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