

CASE REPORT

CARVAJAL SYNDROME: A RARE CARDIOCUTANEOUS SYNDROME

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ABSTRACT: A 3 year old girl was brought by her parents to the DVL OP. Osmania general hospital with the complaints of painful fissures and diffuse thickening of palms and soles since 10 months of her age. She was normal at birth except for presence of woolly hair, thickening of palms and soles started at 10 months of age, which was progressive and lead to formation of fissures. She was put on symptomatic treatment. Carvajal syndrome is very rare disease, so early diagnosis is important to avoid cardiac complications. (Summary)

KEYWORDS: Woolly hair, Palmoplantar keratoderma, Cardiocutaneous syndromes.

INTRODUCTION: Carvajal syndrome also known as 'Striate palmoplantar keratoderma with woolly hair and cardiomyopathy is a cardiocutaneous condition inherited in an autosomal recessive pattern due to a defect in desmoplakin gene. The skin disease presents as a focal non transgradiens 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. It has been described in families from India and Ecuador with rare incidence.⁽¹⁾

CASE REPORT: A 3 years old girl was brought by her parents to the DVL OP with the complaints of painful fissures and diffuse thickening of palms and soles since 10 months of her age.

She was normal at birth except for presence of woolly hair, thickening of palms and soles started at 10 months of age, which was progressive and lead to formation of fissures

No history of Consanguinity among the parents. No history suggestive of Cardio Vascular System involvement.

On Examination: Woolly hair was present (Figure 1). Non transgradient Hyperkeratosis of palms and soles was seen associated with fissuring (Figure 2-3). Keratosis pilaris like lesions over extremities (Figure 4). Nails were dystrophic.(Figure 5) Teeth was normal. Conjunctival, Oral and genital mucosa was normal.

INVESTIGATIONS:

CBP, ESR, RFT, LFT, Sr. Electrolytes, CUE were with in normal limits.

Chest X-ray - normal cardiac and pleuro-pulmonary fields.

ECG -Normal; ECHO -Mild left cardiomyopathy.

Blood samples of parents and patient were sent for the purpose of genetic assessments, reports awaited.

Parents refused biopsy.

As patient ECG didn't show any Arrhythmias, She was not put on any anti arrhythmic medication. Patient was put on symptomatic treatment with emollients and keratolytics. Advised to have regular follow up at Cardiology OP Parents were advised to undergo Genetic Counselling before future pregnancy.

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DISCUSSION: Genodermatoses are inherited genetic skin conditions often grouped into three categories: chromosomal, single gene, and polygenetic. One such example for Genodermatoses is Palmoplantar keratodermas which are a heterogeneous group of disorders characterized by abnormal thickening of the palms and soles. Autosomal recessive and dominant, X-linked, and acquired forms have all been described.

Carvajal syndrome is an example for palmoplantar keratoderma.

Carvajal syndrome is a rare cardiocutaneous syndrome with a triad of woolly hair, striate palmoplantar keratoderma and left ventricular cardiomyopathy.⁽²⁾

It is considered as a variant of Naxos disease with predominantly left ventricular involvement, early morbidity and clinical overlapping with dilated cardiomyopathy which has been described by Carvajal-Huerta et al. from Ecuador.⁽³⁾

Naxos disease is a rare genodermatosis with woolly hair, keratoderma of palms and soles and cardiomyopathy.⁽²⁾ It differs from Carvajal syndrome as it presents with diffuse transgradiens palmoplantar keratoderma and right cardiac abnormalities.

In the Carvajal disease variety two different mutations of the desmoplakin gene (Dsp7901del1G and DspG2375R), affecting the C-terminal of the protein, have been found as causative genes.⁽⁴⁾

Defects in the linking sites of these proteins can interrupt the contiguous chain of cell adhesion, particularly under conditions of increased mechanical stress or stretch, leading to cell death, progressive loss of myocardium and fibro-fatty replacement.⁽⁵⁾

It is a progressive heart disease and may cause sudden death in a child with early age. Presence of syncopal attacks or left ventricular involvement were risk factors for sudden cardiac deaths.⁽⁶⁾

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, anti-arrhythmic drugs and management of heart failure are the recommended treatment modalities.⁽⁷⁾

The population at risk should be genetically screened. And Parents with a child suffering with Carvajal Syndrome should have genetic counselling before any future pregnancy.

Though there is no specific treatment for Carvajal syndrome, symptomatic treatment for painful fissures should be given.

Till now, no case was reported in Osmania General Hospital.

Whenever a child presents with woolly hair associated with any kind of palmoplantar keratoderma, a search for possible cardiac abnormalities is recommended.⁽⁸⁾

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Fig. 1: Woolly Hair



Fig. 2: Hyperkeratosis of Palms



Fig. 3: hyperkeratosis of soles



Fig. 4: Keratosis Pilaris Like Lesions Over Extremities

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Fig. 5: Dystrophic Nails

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