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A clinico-epidemiological study of PLE was done for a period of one year to include 220 cases of PLE of skin type between IV and VI. The manifestation of PLE was most common in house wives on sun exposed areas. Most of the patients of PLE presented with mild symptoms and rash around neck, lower forearms and arms which was aggravated on exposure to sunlight. PLE was more prevalent in the months of March and September and the disease was recurrent in 31.36% of cases.

Comparative study of efficacy and safety of hydroxychloroquine and chloroquine in polymorphic light eruption: A randomized, double-blind, multicentric study

Anil Pareek, Uday Khopkar, S. Sacchidanand, Nitin Chandurkar, Geeta S. Naik 18

In a double-blind randomized, comparative multicentric study evaluating efficacy of antimalarials in polymorphic light eruption, a total of 117 patients of PLE were randomized to receive hydroxychloroquine and chloroquine tablets for a period of 2 months (initial twice daily dose was reduced to once daily after 1 month). A significant reduction in severity scores for burning, itching, and erythema was observed in patients treated with hydroxychloroquine as compared to chloroquine. Hydroxychloroquine was found to be a safe antimalarial in the dosage studied with lesser risk of ocular toxicity.

Many faces of cutaneous leishmaniasis

Arfan Ul Bari, Simeen Ber Rahman

Symptomatic cutaneous leishmaniasis is diverse in its presentation and outcome in a tropical country like Pakistan where the disease is endemic. The study describes the clinical profile and atypical presentations in 41 cases among 718 patients of cutaneous leishmaniasis. Extremity was the most common site of involvement and lupoid cutaneous leishmaniasis was the most common atypical form observed. Authors suggest that clustering of atypical cases in a geographically restricted region could possibly be due to emergence of a new parasite strain.



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Forehead plaque: A cutaneous marker of CNS involvement in tuberous sclerosis

G. Raghu Rama Rao, P. V. Krishna Rao, K. V. T. Gopal, Y. Hari Kishan Kumar, B. V. Ramachandra

In a retrospective study of 15 patients of tuberous sclerosis, eight patients had central nervous system involvement. Among these 8 cases, 7 cases had forehead plaque. This small study suggests that presence of forehead plaque is significantly associated with CNS involvement.

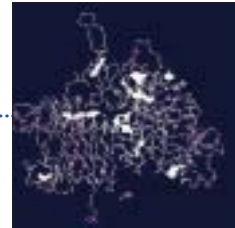


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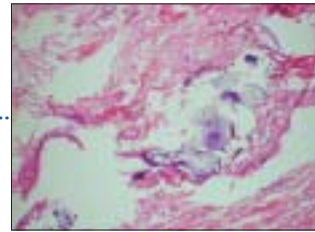
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Naxos disease: A rare occurrence of cardiomyopathy with woolly hair and palmoplantar keratoderma

R. Rai, B. Ramachandran, V. S. Sundaram, G. Rajendren*, C. R. Srinivas

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ABSTRACT

Naxos disease is a rare genodermatosis with woolly hair, keratoderma of palms and soles and cardiomyopathy. A seven-year-old boy presented with woolly hair and hyperkeratotic lesions on the palms and soles since birth. His cardiac status was evaluated and echocardiography revealed early cardiomyopathy. Scalp biopsy revealed hair shaft in an angulated outline suggestive of woolly hair. So the diagnosis of Naxos disease was made. Since he was asymptomatic no treatment was offered but a regular follow-up of the patient and treatment of emergent symptoms should prevent sudden death.

Key Words: Cardiomyopathy, Palmoplantar keratoderma, Woolly hair

INTRODUCTION

Naxos disease is a rare autosomal recessive inherited association of right ventricular dysplasia/dilated cardiomyopathy with woolly hair and palmoplantar keratoderma.^[1] Woolly hair appears from birth, palmoplantar keratoderma develops during the first year of life and cardiomyopathy is clinically manifested by adolescence with 100% penetrance. Patients present with syncope, sustained ventricular tachycardia or sudden death. Symptoms of right heart failure appear during the end stages of the disease. In the Carvajal variant, the cardiomyopathy is clinically manifested during childhood, predominantly involving the left ventricle leading more frequently to heart failure. We report a case of Naxos disease who developed early cardiomyopathy.

CASE REPORT

A seven-year-old boy presented with curly hair from birth and rough palms and soles since the first year of life. On examination, his scalp hair was sparse, fine, pale, curly and

brittle having the appearance of woolly hair [Figure 1]. His palms and soles showed diffuse palmoplantar keratoderma. The keratoderma was more pronounced over the medial aspect of both hands, fingers, heels and forefeet [Figures 2a and b]. He was the second sibling born to consanguineous parents and the previous sibling, had a sudden death. Other members of the family were not affected. His cardiac status was evaluated and electrocardiography showed T inversion in V1-V2. Since he was asymptomatic a Holter monitoring for arrhythmia was not done. A 2D-echocardiography revealed mild but definite dilatation of the right atrium and right ventricle with paradoxical interventricular septum suggesting early cardiomyopathy [Figure 3]. Microscopic examination of the hair with KOH mount did not reveal the presence of fungus. Scalp biopsy revealed epidermis with basket weave keratin. The papillary dermis showed a sparse lymphocytic infiltrate around the blood vessels. Hair shaft showed an angulated outline suggestive of woolly hair. A genetic screening could not be done. Since the patient had right ventricular involvement, woolly hair and palmoplantar keratoderma, a diagnosis of Naxos disease was considered.

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Figure 1: Woolly hair in Naxos disease



Figure 2: (a) Palmar keratoderma, (b) Plantar keratoderma.

DISCUSSION

Naxos disease was first described by Protonotarios *et al.*, in families originating from the Greek island of Naxos. Apart from Naxos, affected families have been detected in other

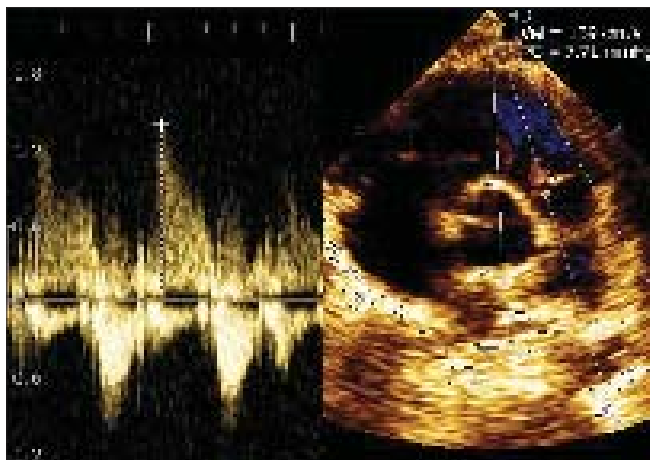


Figure 3: Echocardiogram of the patient showing cardiomyopathy

Greek Aegean islands, Turkey, Israel and Saudi Arabia and two cases have been reported from India.^[2-7] A variety of Naxos disease, reported as Carvajal syndrome presents at a younger age with predominantly left ventricular involvement leading to early heart failure and exhibits a clinical phenotype similar to that of dilated cardiomyopathy.^[2,6]

Woolly hair appears from birth, whereas palmo-plantar keratoderma develops during the first year of life when infants start to use their hands and feet. The cardiomyopathy clinically manifests by adolescence and shows 100% penetrance.^[8] Patients present with syncope, sustained ventricular tachycardia or sudden death. Symptoms of right heart failure appear during the end stages of the disease. One-third patients become symptomatic before the 30th year of life. In some cases, a few clinical findings of an early heart disease can be detected during childhood.

A two base-pair deletion in the plakoglobin (cell adhesion protein) gene (*Pk2157del2TG*), which maps to 17q21, has been identified as the cause of Naxos disease and provided evidence that the pathogenesis might be related to a defect in myocardial mechanical coupling.^[9] 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.^[10] 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.

Naxos disease, associated with progressive cardiomyopathy, has adverse prognosis, especially in the young. In a long-term study of an unselected population of patients with Naxos disease it was shown that risk factors for sudden death

included history of syncope, the appearance of symptoms, severely progressive disease of the right ventricle before the age of 35 years and the involvement of the left ventricle.^[8]

The primary goal is the prevention of sudden cardiac death. Implantation of an automatic cardioverter defibrillator is indicated in patients who develop symptoms and/or structural progression, particularly before the age of 35 years.^[11] Antiarrhythmic drugs are indicated for preventing recurrences of episodes of sustained ventricular tachycardia. In an attempt to control Naxos disease, systematic genetic screening of the populations at risk has been initiated and is starting to identify the heterozygous carriers of the plakoglobin gene mutation.

In this child, the electrocardiography did not reveal any arrhythmia. Since the patient was asymptomatic he was not given any treatment but parents were counseled regarding the disease. A regular follow-up of this patient and treatment at the time of symptoms will prevent sudden death. Naxos syndrome is rare and only two cases have been reported from India. Although cardiomyopathy usually manifests by adolescence, this patient had early involvement of his right ventricle.

Whenever woolly hair is associated with any kind of palmoplantar keratoderma, a search for possible cardiac abnormalities is recommended.^[2]

REFERENCES

1. Protonotarios N, Tsatsopoulou A, Patsourakos P, Alexopoulos D, Gezerlis P, Simitsis S, *et al.* Cardiac abnormalities in familial palmoplantar keratosis. *Br Heart J* 1986;56:321-6.

2. Carvajal-Huerta L. Epidermolytic palmoplantar keratoderma with woolly hair and dilated cardiomyopathy. *J Am Acad Dermatol* 1998;39:418-21.
3. Barker JN, Protonotarios N, Tsatopoulou A, MacDonald DM. Palmoplantar keratoderma, curly hair and endomyocardial fibrodysplasia: A new syndrome. *Br J Derm* 1988;119:13-4.
4. Tosti A, Misciali C, Piraccini BA, Fanti PA, Barbareschi M, Ferretti RM. Woolly hair, palmoplantar keratoderma and cardiac abnormalities: Report of a family. *Arch Dermatol* 1994;130 522-4.
5. Bukhari I, Juma'a N. Naxos disease in Saudi Arabia. *J Eur Acad Dermatol Venereol* 2004;18:614-6.
6. Rao BH, Reddy IS, Chandra KS. Familial occurrence of a rare combination of dilated cardiomyopathy with palmo plantar keratoderma and curly hair. *Indian Heart J* 1996;48:161-2.
7. Adhisivam B, Mahadevan S. Naxos disease. *Indian J Pediatr* 2006;73:359-60.
8. Protonotarios N, Tsatopoulou A, Anastasakis A, Sevdalis E, McKoy G, Stratos K, *et al.* Genotype-phenotype assessment in autosomal recessive arrhythmogenic right ventricular cardiomyopathy (Naxos disease) caused by a deletion in plakoglobin. *J Am Coll Cardiol* 2001;38:1477-84.
9. McKoy G, Protonotarios N, Crosby A, Tsatsopoulou A, Anastasakis A, Coonar A, *et al.* Identification of a deletion in plakoglobin in arrhythmogenic right ventricular cardiomyopathy with palmoplantar keratoderma and woolly hair (Naxos disease). *Lancet* 2000;355:2119-24.
10. Norgett EE, Hatsell SJ, Carvajal-Huerta L, Cabezas JC, Common J, Purkis PE, *et al.* Recessive mutation in desmoplakin disrupts desmoplakin-intermediate filament interactions and causes dilated cardiomyopathy, woolly hair and keratoderma. *Hum Molec Genet* 2000;9:2761-6.
11. Gatzoulis K, Protonotarios N, Anastasakis A, Tsatsopoulou A, Vlasseros J, Gialafos J, *et al.* Implantable defibrillator therapy in Naxos disease. *Pacing Clin Electrophysiol* 2000;23: 1176-8.