

Case Report

Clouston's syndrome: a rare case report

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Received: 14 July 2020

Accepted: 06 August 2020

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ABSTRACT

Ectodermal dysplasias are a heterogeneous group of disorders with primary defect in hair, teeth, nail and sweat glands with an estimated frequency of about seven per 10,000 births. Numerous types have been described and several classifications exist. Clouston's syndrome (hidrotic ectodermal dysplasia) is a rare genodermatosis, characterized by a triad of nail dystrophy, alopecia and palmoplantar hyperkeratosis. Clouston's syndrome is transmitted as an autosomal dominant trait and caused by mutations in the GJB6 gene (13q12), encoding the gap junction protein connexin 30 (CX30). At present, there is no treatment for the disease and management is purely supportive. The improved prognosis over time is likely due to greater recognition of the condition. In this report, a 23-year-old male patient with nail abnormalities and thickening of palmoplantar skin is reported. Anodontia of permanent dentition was present along with androgenic alopecia.

Keywords: Clouston's syndrome, Alopecia, Palmoplantar hyperkeratosis, Nail dystrophy

INTRODUCTION

Thuram published the first report of patient with ectodermal dysplasia in 1848, but the term ectodermal dysplasia (ED) was not coined until weech termed it as so in 1929.^{1,2}

The ectodermal dysplasias are a large group of hereditary disorders characterized by alteration of structures of ectodermal origin. They are congenital, diffuse and non-progressive disorders. They are categorized into two main groups: X-linked recessive anhidrotic (Christ-Siemens-Touraine syndrome) and hidrotic ectodermal dysplasias (Clouston's syndrome)³.

Clouston's syndrome is characterized by the triad of nail dystrophy, palmoplantar hyperkeratosis and generalized hypotrichosis. We report a case of Clouston's syndrome associated with Androgenic Alopecia.

CASE REPORT

A 23-year-old male patient reported to our clinic with a chief complaint of thickening of palmoplantar skin, which had an insidious onset with gradual progression. Dry skin over the face was present since birth. His medical history was non-contributory and he was born out of consanguineous marriage with normal vaginal delivery. Patient's mother did not suffer from any disease during pregnancy and two of his siblings died of unknown causes. Detailed history revealed that there was spontaneous shedding of teeth one year back with decreased sweating.

Physical examination revealed thickened, striated, and discoloured finger nails (Figure 1) and hyperkeratosis of palms and soles (Figure 2). Frontal bossing, prominent supraorbital ridge, sunken cheeks, sparse hair over the body, low-set over-folding ears, and conical teeth (Figure 3) and androgenic alopecia (Figure 4), were noted.



Figure 1: Dystrophic nails with striations.



Figure 2: Hyperkeratosis of palms and soles.



Figure 3: Conical or peg shaped teeth.



Figure 4: Androgenic alopecia.

Other cutaneous and systemic examinations were normal. Routine investigations like Complete blood count, Blood sugar, Liver Function Tests, Renal Function Tests, electrocardiogram, Chest X-ray, Abdominal ultrasound

and Urine routine were normal. Orthopantogram revealed peg shaped teeth with partial anodontia (Figure 5). Skin biopsy revealed normal sweat glands.

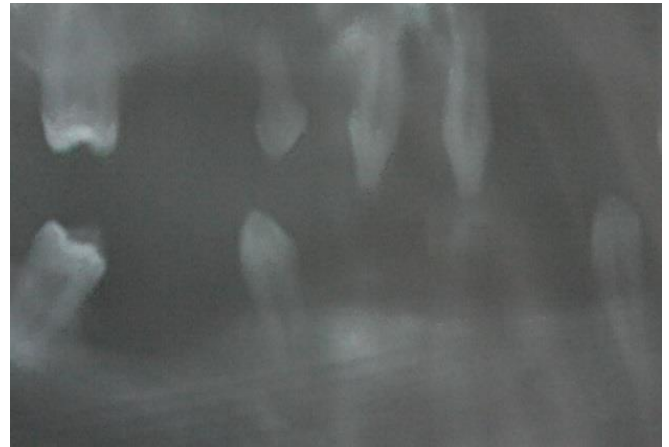


Figure 5: Orthopantogram showing peg shaped teeth and missing teeth.

Based on clinical and histological features a diagnosis of hidrotic ectodermal dysplasia was made. Patient was counselled about the nature of the disease, proper skin care, and regular follow up and was referred to a specialized dental rehabilitation centre for orthodontic treatment and prosthetic rehabilitation.

DISCUSSION

The ED are a heterogeneous group of hereditary disorders which occur approximately one in every 100,000 births which are caused by primary developmental defects of two or more embryonic ectoderm derived tissues.⁴

Classification of EDs based on clinical findings is more relevant and may be divided into two broad categories. Hypohidrotic ectodermal dysplasia and hidrotic ectodermal dysplasia.^{5,6}

Clouston's syndrome was first described in 1895 and later reported in Canadian families.⁷ It is characterized by the triad of nail dystrophy, generalized hypotrichosis, and palmoplantar hyperkeratosis. Sparse hair and nail dystrophy are present since one month of life. Progressive loss of hair may lead to total alopecia at puberty. Nails gradually become dystrophic during childhood. Nail clubbing may occur. Palmoplantar keratoderma may develop in childhood and progress with age. Clinical features vary greatly among individuals, even within the same family.⁸

In addition, some patients may have hyperpigmentation, more evident on the joints. Strabismus, conjunctivitis, cataracts, deafness, polydactyly and syndactyly may occur.⁸ Eccrine syringe fibroadenomas have been reported in some patients.⁹ Epidermal cysts were

documented recently.¹⁰ This condition primarily affects hair and nails. Sweating is normal.

Diagnosis/testing hidrotic ectodermal dysplasia 2 is suspected after infancy on the basis of clinical features in most affected individuals. GJB6 is the only gene known to be associated with Hidrotic ectodermal dysplasia 2. Targeted mutation analysis for the four most common GJB6 mutations is available on a clinical basis and detects mutations in approximately 100% of affected individuals.¹²

There is no treatment for this disorder and management is purely supportive. Management of patients with this condition usually includes a removable and/ or fixed partial denture, complete denture prosthesis, when indicated. These treatment approaches can be used individually or in combination to provide an optimal result.

Special hair care products to help manage dry and sparse hair, wigs, emollients to relieve palmoplantar hyperkeratosis.⁸

CONCLUSION

To conclude, we must think of the possibility of Clouston's syndrome even if the patient has presented with nail dystrophy. This report might help physicians and dermatologists not to miss the diagnosis of Clouston's syndrome even in cases presenting merely with nail dystrophies as seen in our patient.

Funding: No funding sources

Conflict of interest: None declared

Ethical approval: Not required

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Cite this article as: Karjigi S, Reddy AG, Herakal K, Reddy V. Clouston's syndrome: a rare case report. Int J Res Dermatol 2020;6:702-4.