Case Report

Clouston's Syndrome-A Case Report

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Abstract

A heterogeneous group of disorders namely Clouston's syndromes characterized by a primary defect in teeth, hair, nail and sweat glands with an estimated frequency of about seven per 10,000 births. Hidrotic ectodermal dysplasia or Clouston syndrome is a rare inherited disorder of ectodermal dysplasia. It is an autosomal-dominant disease and is identified by a triad of palmoplantar keratoderma, alopecia and nail dystrophy. 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 3-year-old child with nail abnormalities and painful thickening & fissuring of palmoplantar skin is reported.

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Introduction

Clouston syndrome is an autosomal dominant inherited disease.[1] It is distinguished by the clinical triad of alopecia, palmoplantar hyperkeratosis and nail dystrophy. This syndome was first described in the Canadian population. It is caused due to GJ-B6 gene mutation located on chromosome 13q11-12 which codes for connexin 30(Cx30).[2] The patients from the same family who are affected with this dis-ease show variability in expression but the disease penetrance is complete.[3] In infants the scalp hair are brittle, wiry, pale and patchy. During puberty, total alopecia may occur because of progressive hair loss from the scalp and other body parts. Nails may appear milky white in early childhood in these patients. In the mildest condition it was noticed that the nails may merely be thickened and slightly discolorured, striated longitudinally, and, if long, bent forward at the finger-tip like a beginning claw. Nails be-come thick, short, separated from the nail bed distally and eventually become dystrophic.[4] There is slow nail growth. The severity of palmoplantar ke-ratoderma increases with age.[3] The degree of skin alteration does not run absolutely parallel with the severity of the hair and nail dystrophy. It is more marked in the males than in the females and more in the adult than in the children.[4] Dental findings include hypodontia, microdontia and sometimes anodontia along with lack of alveolar ridge.[5] There is often increased pigmentary changes over the phalangeal joints and also on the elbows and knees. There can be pigmentation in the axilla and the genitals. Ocular features include strabismus, pterygium, conjuncttivitis, premature cataracts and mental development may be retarded. Endocrine involvement may cause features like pigmentation of skin

over joints, thickened skull bones, sausage-shaped digits.

There is no specific treatment for this syndrome. Keratolytics and emollients are used for palmoplantar keratoderma. Professional pedicures and manicures can reduce obvious aspects of nail changes. Tretinoin which enhances the absorption of minoxidil can be effective in hidrotic ectodermal dysplasia.[6]

Case Report

A child who was 3 years old was brought to skin opd, Mayo Hospital, Lahore by her parents with chief complaints of sparse scalp hair with progressive hair loss since infancy, thickened and dystrophic nails since 1 year and painful fissuring of palms and soles since 1 year. The child was born to a consanguineous couple via normal vaginal delivery at home and her birth weight was normal. The baby cried immediately after delivery.

On examination, there were sparse scalp hairs which were twisted, brittle and pale. Eyelashes were short and sparse and eyebrows were absent. Nails were thickened with subungual debris, yellowish coloured nail plates and some nails were dystrophic. There was palmoplantar thickening with fissuring of palms and soles.

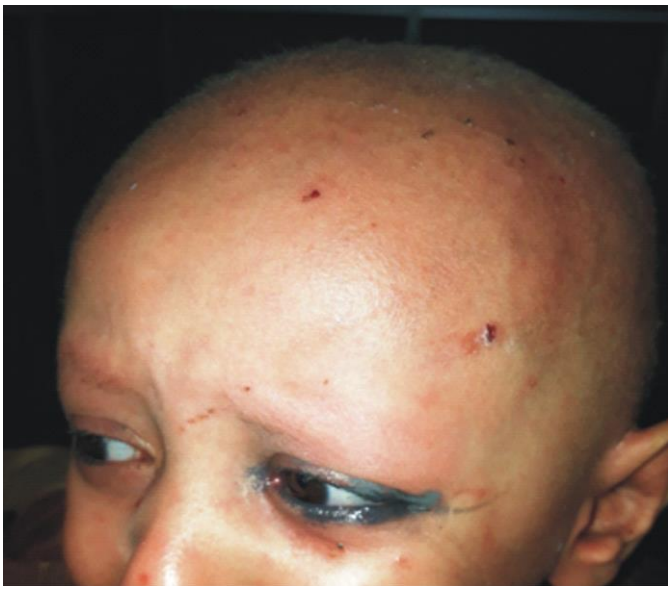

Figure 1: *Sparse, twisted, brittle and pale scalp hair. Loss of eyebrows, sparse and short eyelashes*

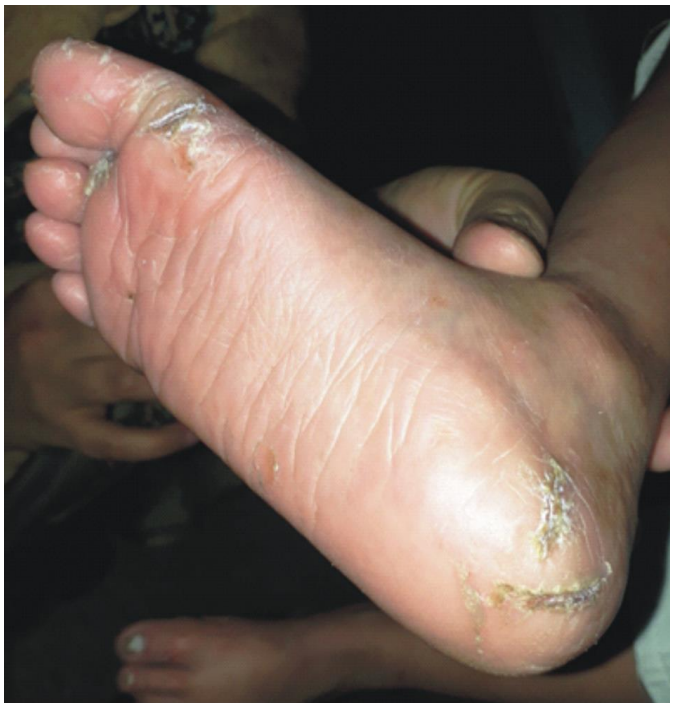

Figure 2: *Thickened sole of feet with fissuring*

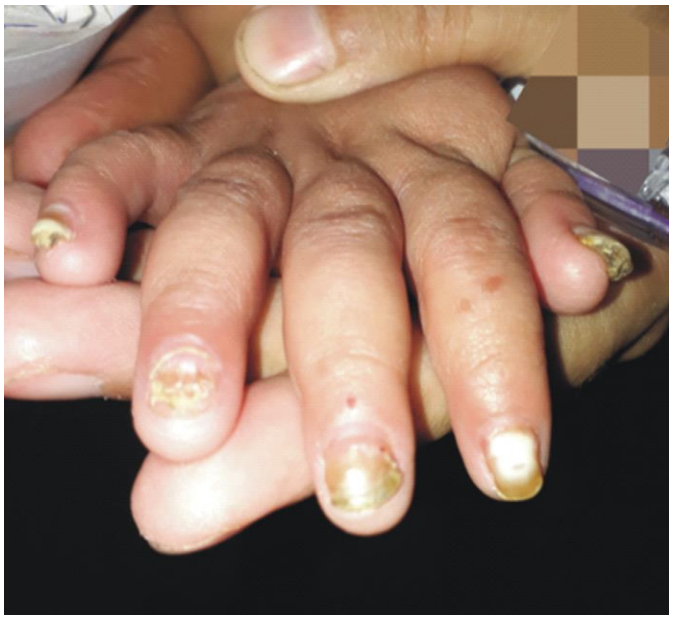

Figure 3: *Thickened dystrophic yellowish nails*

Discussion:

Clouston syndrome is identified by abnormalities of the hair, skin and nails. It was first described in 1895.[7] In this there is nail dystrophy, palmoplantar hyperkeratosis and generalized hypotrichosis, Hair are sparse and dystrophy of the nails are also prominent features. Total alopecia may occur at puberty due to progressive hair loss. Gradually, nail dystrophy and nail clubbing occur during childhood.

Gradual progression of palmoplantar keratoderma with increasing age. Variation in the clinical features may occur among individuals and within the same family me-mbers.[8] Some patients may develop hyperpigmentation which is more evident over the joints. In addition, conjunctivitis, cataracts, strabismus, deafness, syndactyly & polydactyly may occur.[8] Eccrine fibroadenomas and epidermal cysts may also occur.[9,10] This condition primarily affects hair and nails. Sweating is normal. Diagnosis of hidrotic ectodermal dysplasia 2 is suspected when there are evident clinical features in majority of affected individuals. The only gene associated with Hidrotic ectodermal dysplasia 2 is GJB6. Mutation analysis of the GJB6 gene mutation detects the mutations of four types in 100% of the affected individuals of Clouston's syndrome.[10]

Singh et al[4] reported a 9 year old child with Clouston's Syndrome with palmoplantar keratoderma. In this case anodontia was present but there was no alopecia. Andrade et al[8] described a patient with Clouston's Syndrome, who presented with papulonodular lesions in the extremities which were compatible with eccrine syringofibroadenoma both clinically and histologically.

There is no treatment for this disorder and management is purely supportive. Multidisciplinary approach is required. Complete denture prosthesis, fixed/removable partial denture may be required in these patients according to the indication. Dry and sparse hair can be managed by hair care medications. For palmoplantar hyperkeratosis emollients and keratolytics can be used. All the treatment options are either used in combination or individually depending upon the clinical presentation of the patient to get maximum benefit.

Conclusion:

Possibility of Clouston's syndrome should be kept in mind even if the patient presented with nail dystrophy. This report might help physicians and dermatologists not to miss the diagnosis of Clouston's syndrome in cases presenting with alopecia, nail dystrophies and palmoplantar fissuring.

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