CASE REPORTS

SIEMENS SYNDROME

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A 10-year-old girl born of a consanguinous marriage presented with localised plantar keratodermas associated with deafness, physical and mental retradation. Although thiis disease has autosomal dominant mode of inheritance, no family history of similar illness was seen in our case. And also physical and mental retardation were not reported in literature as associated features.

key word : Siemens syndrome

Introduction

A large group of localised keratodermas has been distinguished principally on morphological grounds, comprising areata, striata and punctate keratodermas. There are demonstrable genetic differences between many of these disorders but differentiation based on morphology alone has led to voluminous and unrewarding leterature.[1] Keratosis palmoplantaris striata is known as Siemens syndrome.[2] Herewith we report a case of Siemens, syndrome for its rarity.

Case Report

A 10-year-old girl born of cons - anguinous marriage present with raised skin lesion over planatar aspect of the feet since early childhood. There was history of physical and mental ratardation and deafness since infancy. There was no involvement of hair or teeth. And also there was no history of similar illness in the family.

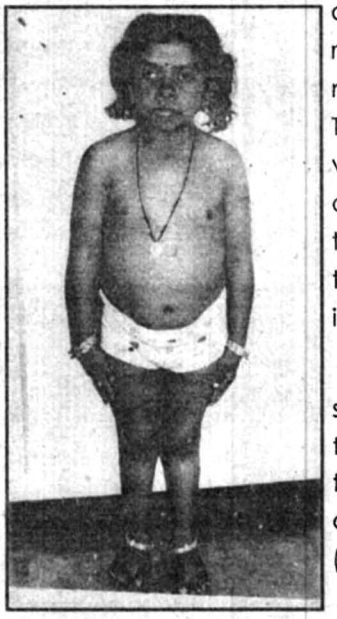

Fig - 1. Clinical photograph showing physical retardation

General and systemic examination revealed mental as well as physical retardation (Fig.1). Cutaneous examination revealed welll - defined islands of multiple hyperkeratotic lesions over the soles (Fig. 2).The palms were normal. Hair, teeth, and nails were also normal.

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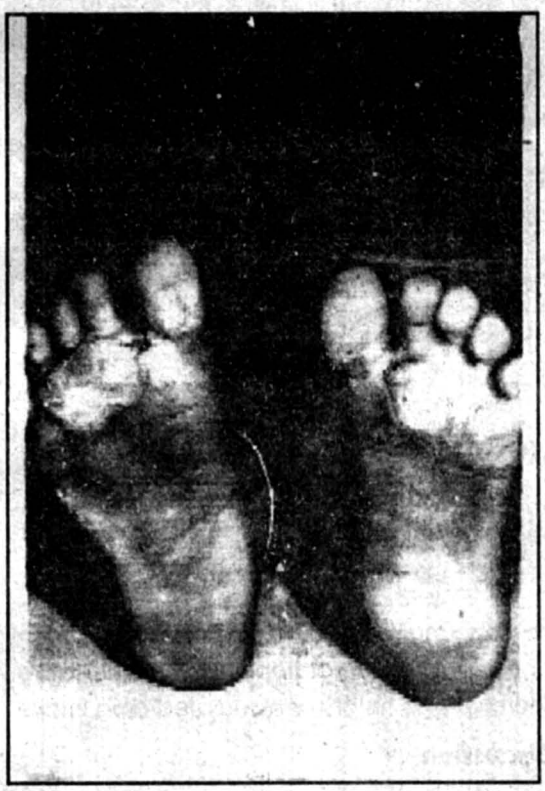

Fig - 2. localised plantar keratoderma

Routine blood and urine investigations were within normal limits. Serum calcium, phosphorous, TSH, T_3 and T_4 levels were also within normal limits. Skeletal survey showed only minimal osteoporosis. Audiogram showed bilateral mild conductive hearing loss. Based on the clinical and laboratory findings, the patient was diagnosed as Siemens syndrome.

Discussion

Siemens syndrome is inherited as an autosomal dominant. The earliest sign is palmar or plantar erythema followed by islands or linear hyperkeratotic lesions.[3] Associated features commonly encountered are teeth enamel dysplasia, total leuconychia, steely hair and deafness.[4] But in our case deafness was associated with physical as well as mental retardation.

References

1. Griffiths WAD, Leigh IM, Marks R. Disorder of keratinisation. In Texbook of Dermatology Champion RH, BurtenJI, Ebling FJG, eds, 5th Edition, Oxford: Blackwell Scientific Publications, 1992 ; 1325 - 1390.

2. Siemens H W. Keratosis palmoplantaris striata. Arch Dermatol Syphilol 1929 ; 157 : 392 - 408.

3. Kientz T. Keratosis palmoplantaris striata, Brunnauer Fuhs 1927 (Keratosis palmoplantaris various wachtterss 1963). Hautarzt 1976;27:403.

4. Crosti C, Sala F, Leuconychic totale et dysplasia ectodermique ; observation de deur cas. Ann Dermatol Venereol 1983 ; 110 : 617 - 622.