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ANGIOKERATOMA CORPORIS DIFFUSUM

R P Sharma, N K Sharma, S Mithal, A K Agrawal

A case of angiokeratoma corporis diffusum having some classical features with dilatation of retinal vessels, but without urinary signs of renal involvement, is reported.

Key Words : Fabry-Anderson disease, Corneal verticillata, Acroparaesthesia

Introduction

In 1898 Fabry and Anderson independently described a form of angiokeratoma now known as angiokeratoma corporis diffusum.[1] It is a rare X-linked recessive disorder. Deficiency of lysosomal hydrolase alpha-galactosidase-A results in the progressive deposition of uncleaved neutral glycosphingolipids - predominantly alpha-galactosyl-lactosyl ceramide (trihexosyl ceramide) within the lysosomes of endothelial, perithelial and smooth muscle cells.[2] The gene responsible for expressing alpha-galactosidase-A has been localised to the middle of the long arm of X-chromosome.[3] Most cases occur in males and females are usually symptomless carriers. The cutaneous eruptions usually first appear shortly before puberty. Neural pains are usually presenting symptoms during childhood while vasomotor disturbances usually appear at a later stage. Eye lesions like corneal dystrophy and dilatation of conjunctival and retinal vessels may be present.[4] Patients usually die in third or fourth decade from vascular accident or uraemia.

Case Report

An 18-year-old male patient complained of generalised dryness and hypohidrosis for last 8 years and gradual development of reddish brown eruptions on abdomen, back, buttocks, genitals and thighs. The initial lesions started around umbilicus. The lesions subsequently became numerous and prominent. He also complained of some degree of distress over hands and feet and recurrent erythematous lesions for last three years. History of consanguinity or similar disease in family was absent. General examination revealed dull and vacant look of face, the anterior hair line was low, lips were large and thick, front teeth were widely spaced and gums were swollen (Fig. 1). Skin was dry and studded with multiple brownish papules along with erythematous macules (Fig. 2). Lesions were maximally present on back, around umbilicus, genitals and thighs. At places lesions were hyperkeratotic. No specific mucosal lesions were present. Scalp hair were coarse but had normal texture and

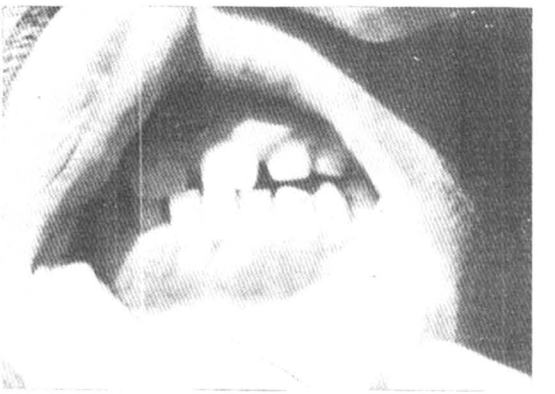

Fig. 1. Thick lips, widely spaced teeth and swollen gums.

From the Departments of Dermatology, Ophthalmology and Pathology, LLRM Medical College, Meerut - 250 004, India.

Address correspondence to : Dr R P Sharma

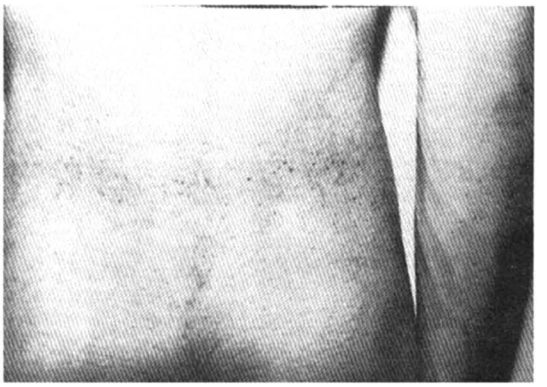

Fig. 2. Multiple papular lesions on back.

pattern. Nails were deformed and had turtle back convexity. Eye examination showed corneal verticillata and fluorescin angiography revealed dilated retinal vessels

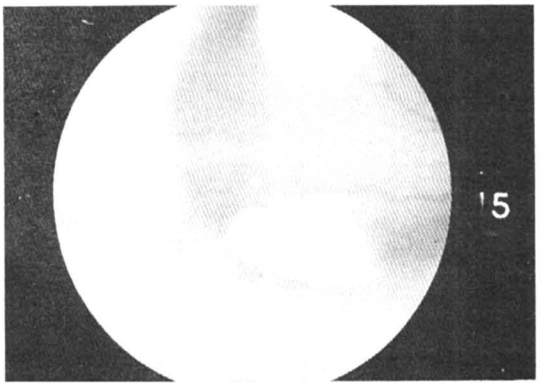

Fig. 3. Dilated retinal vessels.

(Fig. 3). His mother was also found to have corneal verticillata. Examination of other systems and autonomic function tests were within normal limits.

Routine and specific blood tests were within normal limits. Routine urine examination did not show any abnormality. Examination of urinary sediment for birefringent lipid containing cells was positive. Specific tests to detect alpha-galactosidase- A deficiency and presence of ceramide trihexoside in urine could not be done due to lack of facilities. Histopathology of the cutaneous lesions was compatible with clinical diagnosis of angiokeratoma corporis diffusum.

Discussion

The present case had some classical features such as generalised dryness, hypohidrosis, dull and vacant facial appearance, acroparaesthesia, angiokeratomas, corneal verticillata and gingivitis. We could find only one Indian reference of a case reported by Sharma et al.[5] Present case had many findings similar to their case except few features like complete absence of sweating, stooping posture and renal involvement etc. In addition present case had dilated and tortuous retinal vessels. We could not find any urinary sign of renal involvement in present case.

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