DYSKERATOSIS CONGENITA ASSOCIATED WITH 'EMPTY' SELLA

RM Govalkar*, A Parthasaradhi*, SK Wangnoo**

A case of dyskeratosis congenita (DC) associated with 'empty' sella in a 22-year old Arab male born of consanguineous marriage is reported. He exhibited all the essential features of DC and various ocular and dental abnormalities. In addition, lateral radiograph of the skull revealed enlargement of the pituitary fossa and cranial CT scan showed features suggestive of 'empty' sella. The association of 'empty' sella with dyskeratosis congenita does not seem to have been recorded in earlier literature.

Key Words : Dyskeratosis congenita, 'Empty' sella

Introduction

Dyskeratosis congenita (DC) is a rare hereditary disorder with classical triad of reticulate pigmentation, nail dystrophy and leukoplakia of the mucous membranes. Variety of other features like Fanconi type pancytopenia, ocular and dental abnormalities and immunological defects have been described. Abnormalities of the CNS like low intelligence, small sella turcica and intracranial calcifications have also been reported. Our patient was found to be mentally sound and the radiograph of the skull revealed enlarged sella turcica. The cranial CT scan showed features suggestive of 'empty' sella, which was an unusual feature in the present case.

Case Report

A 22-year-old Arab male born of consanguineous marriage presented with progressive cutaneous and nail changes since the age of 6 years. Skin changes in the form of scaling especially over th dorsa of both hands were noticed, whic progressed to cause atrophy of the ski Spontaneous pigmentary change developed initially over the abdomen an subsequently involved lower back, thigh and forearms. The nail changes wer noticed at about 6 to 8 years of age an gradually within a few years nails of a digits were completely destroyed. Ther was no history of any periungu infection, hyperhidrosis of palms an soles, bullous eruptions o photosensitivity. He had excessiv lacrimation and recurrent conjunctivitis o both eyes since the age of 10 years There were no urinary complaints o bowel irregularities. There was no histor of dysphagia, any blood transfusions i the past. His general health had bee excellent. He had no mental difficulties

The patient was hyposthenic wit normal vital parameters. The pigmenta changes included brownish-black discre macules of 2-3 mm size over the butter area of the face and grayish-brow mottled pigmentation involving lowe trunk and thighs over an apparentl normal skin. The reticula hyperpigmentation with hypopigmente

From the Departments of Dermatology,* and Medicine,** Hail General Hospital, Hail, Saudi Arabia.

Address correspondence to :
Dr Ramesh M Govalkar,
P.O. Box : 2154, Hail Kingdom of Saudi Arabia.

atrophic macules and telangiectasia were present over the flexor aspect of both forearms. The skin over the dorsa of the hands (Fig. 1) and feet, elbows and knees was atrophic, shiny, translucent and wrinkled. The palms and soles were erythematous, shiny and atrophic showing loss of dermatoglyphic pattern over the fingertips, without any sensory deficit. The scrotal skin was atrophic and there was scaling over the glans penis. The testes were of normal size.

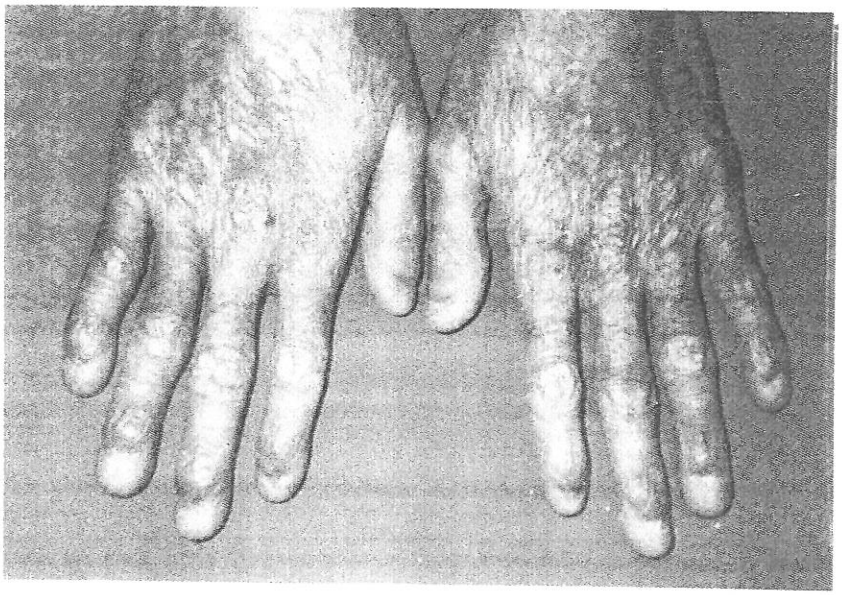

Fig. 1. Atrophic wrinkled skin over the dorsa of both hands with complete dystrophy of nails

The nails (Fig. 1) of all digits were completely destroyed and were replaced by an atrophic and fibrotic skin. The proximal and lateral nail folds were absent. The oral cavity showed persistent firmly adherent irregular white patches of leukoplakia on the dorsa of the tongue (Fig. 2) and loss of papillae over the margins and tip of the tongue. The buccal mucosa was normal. The dental examination revealed chronic gingivitis, periodontitis, wide spacing, rotation and mal-alignment of teeth, extensive caries and loss of few teeth of both the jaws. The slit lamp examination of eyes showed bilateral atresia of lacrimal puncti of both upper and lower eyelids. The visual acuity, visual fields and fundus examination were normal. Sparsness of the ciliary hair and premature canities were also present. Anorectal mucosa was normal.

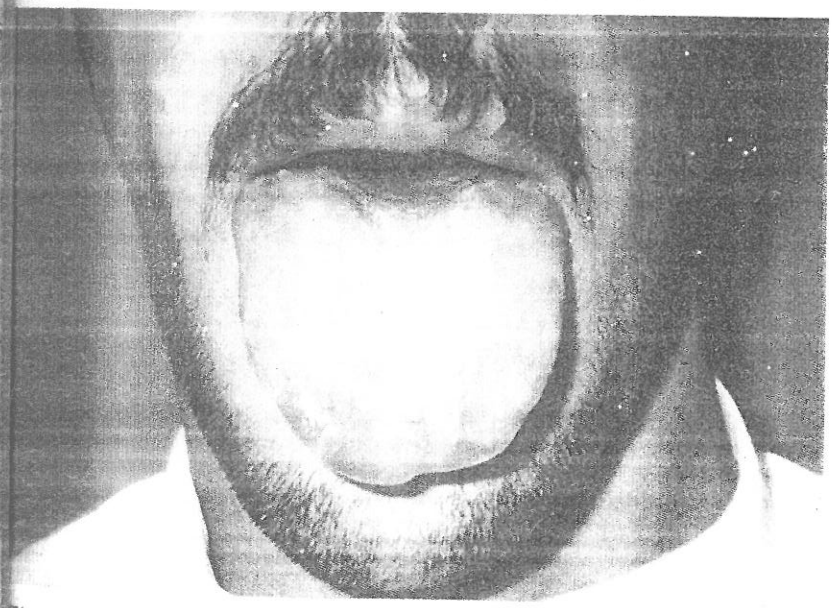

Fig. 2. Leukoplakia on the dorsa of the tongue with loss of lingual papillae at the margins

The parents and other siblings (3males, 1 female) did not have similar abnormalities.

The routine laboratory tests were negative or within normal limits. Histopathology of the reticulate pigmented skin on the forearms showed epidermal atrophy, focal degeneration of basal cell layer, melanin incontinence in upper dermis and mononuclear perivascular infiltrate in the upper and mid-dermis. Radiological studies of the chest, hands and feet, long

bones and paranasal sinuses showed no abnormalities. The lateral X-ray of the skull showed enlarged pituitary fossa with no bony erosions. In the sagittal plane

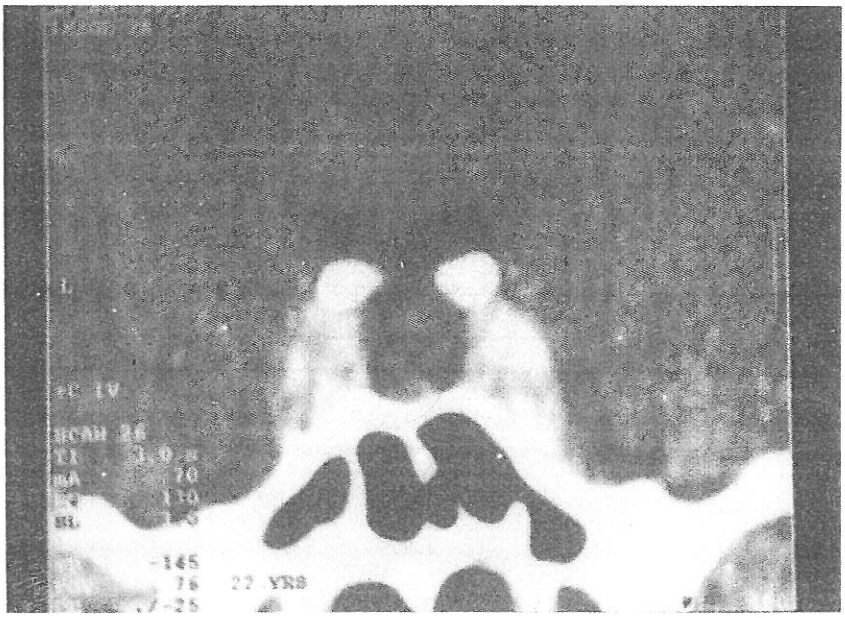

Fig. 3. Coronal CT scan showing hypodense intrasellar area

the length and depth of the pituitary fossa were 19 mm and 15 mm respectively (normal maximum values: length 17 mm and depth 14 mm). The cranial CT scan displayed hypodense intrasellar area (Fig.3) with no evidence of abnormal intravenous enhancement and was suggestive of 'empty' sella. There was no evidence of intracranial calcifications.

Comments

The diagnosis of dyskeratosis congenita in our patient was supported by the presence of characteristic triad of pigmentary and atrophic changes of the skin, nail dystrophy and leukoplakia on the tongue. In addition, he had variety of minor manifestations including dental and ocular abnormalities as have been described in earlier reports. There appears to be genetic heterogeneity in DC. The usual mode of inheritance is an X-linked recessive, with the majority of cases occurring in males born [illegible] nonconsanguineous marriage. Out [illegible] approximately 108 reported cases abo[illegible] 7 cases have been reported resulting fro[illegible] 4 consanguineous marriages. In 5 of these cases th[illegible] inheritance was an autosom[illegible] dominant type.[2] Our patie[illegible] was born of consanguineo[illegible] marriage and this seems to b[illegible] the eighth reported ca[illegible] resulting from the fif[illegible] consanguineous marriage. Th[illegible] mode of inheritance is no[illegible] clear.

We observed an unus[illegible] distribution of mottled grayis[illegible] brown pigmentation overlyin[illegible] an apparently normal ski[illegible] which was more pronounce[illegible] over the lower trunk and thighs. Unlik[illegible] previous cases the reticulate pigmentatio[illegible] resembling poikiloderma clinically an[illegible] histopathologically was present only o[illegible] the flexor aspect of the forearms in[illegible] linear pattern sparing the usual site[illegible] (face, neck and upper trunk).

Enlargement of pituitary fossa in o[illegible] case is in contrast th the earlier reports[illegible] small sella turcica described in DC.[4] Th[illegible] normal endocrine functions, absence[illegible] visual field defects, normal visual acui[illegible] and normal fundi in our patient ruled o[illegible] the possiblity of intrasellar tumour[illegible] raised intracranial tension as the cause[illegible] enlarged sella turcica. Further studies wi[illegible] CT scan of cranium revealed hypoden[illegible] intrasellar area without any evidence[illegible] abnormal intravenous enhancemen[illegible] which was suggestive of presence of C[illegible] in the sella. Negative history of any sk[illegible] surgery, the positive findings in skull

ray and CT scan, normal hormonal profile were all correlating well with the diagnosis of primary 'empty' sella.

In primary 'empty' sella the suprasellar subarachnoid recess filled with CSF herniates into the pituitary fossa, through the developmental defect in the diaphragma sella and the fluid transmitted pulsations result in enlargement of the sella turcica.[5]

We do not know whether primary 'empty' sella is one of the many abnormalities associated with DC or a chance simultaneous occurrence. The occurrence of enlarged sella turcica with 'empty' sella does not seem to have been recorded in earlier cases. Both these conditions being ectodermal developmental disorders, we assume that primary 'empty' sella is one of the many abnormalities associated with dyskeratosis congenita.

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