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# ATYPICAL GORLIN'S SYNDROME

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A 21-year-old woman presented with complaints of skin lesions on her face, palms and soles. On examination, 8 to 10 well-defined, pigmented nodules with raised and pearly borders were seen on the face. Multiple pits were present on the palms and soles. Biopsy of both lesions revealed the presence of basal cell epitheliomas. The patient also gave history of removal of a keratocyst of mandible 10 years back. The case was diagnosed as Gorlin's syndrome. It is atypical due to the low number of basal cell epitheliomas, the occurrence of only a single odontogenic cyst and the absence of other features usually associated with this condition.

**Key Words : Gorlin's syndrome, Atypical basal cell epithelioma**

## Introduction

Basal cell epitheliomas are common tumours which can occur alone or as part of a syndrome. Three clinical syndromes in which basal cell epitheliomas play an important part include (1) the naevoid basal cell epithelioma syndrome, (2) the linear unilateral basal cell naevus, and (3) the Bazex syndrome. The following case report is an atypical presentation of naevoid basal cell epithelioma syndrome or Gorlin's syndrome. This entitiy is a genetically determined disorder transmitted in an autosomal dominant fashion with multiple and variable effects.[1] It is characterised by the occurrence of multiple basal cell epitheliomas early in life, pittting of the palms and soles,[2] odontogenic cysts of the jaw, various skeletal defects and central nervous system disturbances.

## Case Report

A 21-year-old woman presented with complaints of skin lesions in her face, palms and soles since childhood. She first noted pits in the palms and soles which were asymptomatic and were increasing in number over the past few years. On examination, there were multiple, well defined pits in the palms and soles (Figs.

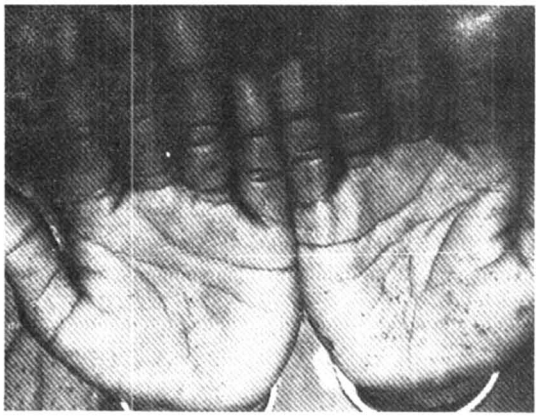

Fig .1. Palms showing multiple pits.

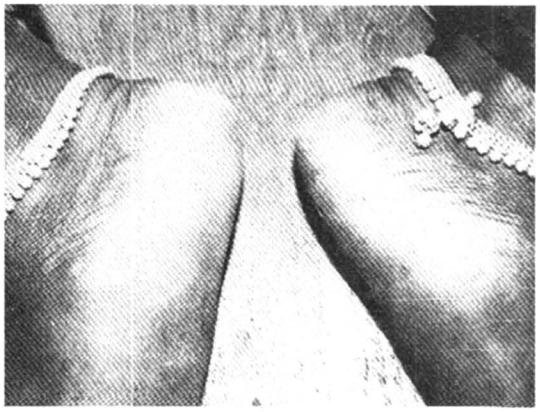

Fig .2. Multiple well-defined pits over the soles.

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1,2). There were also pigmented, well defined, discrete, painless, mobile, non-tender nodules, about 8-10 in number and present over the face below the lower eyelid on both sides (Fig. 3) and post-auricular region of the right ear.

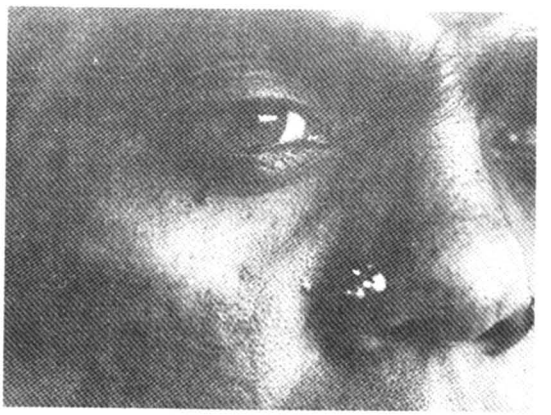

Fig .3. Tiny, pigmented papular lesion just below the right infraorbital region.

The borders were raised and pearly . Her hair, nails and mucosa were normal. In addition, the patient gave history that 10 years ago, she underwent surgery for a swelling in the jaw. The histopathology report was one of a keratocyst of the mandible. There was no family history of similar lesions.

Routine investigations were within normal limits. X-ray of the jaw revealed no abnormality. Biopsies were performed on three of the lesions of the face. All showed features of basal cell carcinoma (Fig. 4). Biopsy of one of the palmar pits also revealed findings compatible with basal cell epithelioma.

## Discussion

In any genodermatosis, pleiotropism may be expected with variable clinical manifestations of a single genetic aberration. Gorlin's syndrome is no exception to this fact as demonstrated by the atypical presentation of our case.

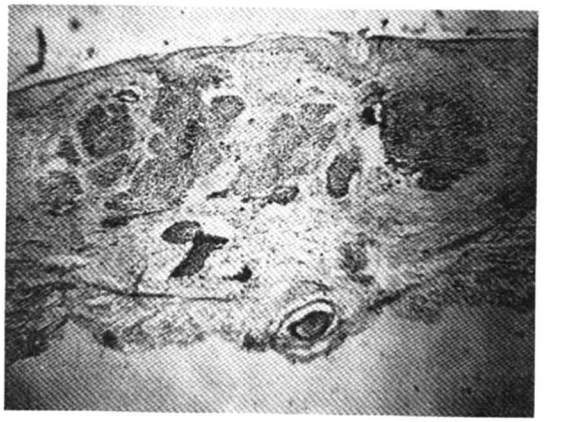

Fig .4. Photomicrograph showing islands of basilioma cells with peripheral clefts (H&E x 60).

The skin manifestations of multiple basal cell carcinomas in our case started in childhood and numbered between 8 and 10. This contrasts with the classical presentation in which the tumours are innumerable and distributed in a bilateral and symmetrical manner.[2] Other cutaneous manifestations of the syndrome include milia, epidermal and sebacious cysts, lipomas and fibromas, none of which were observed in our case.

The dental cysts occurring in Gorlin's syndrome are usually multiple, occurring in either jaw or both. The mandible is more often involved than the maxilla. Histopathologically, they are odontogenic keratocysts.[3] They are the most common stigma of the syndrome. Other skeletal abnormailites may also be present.

Pits of the hands and feet are characteristic features of the syndrome.[2] They have their onset during the second decade and become more frequent with advancing age. Our case showed these characterişitics and in addition showed the presence of small basal cell epitheliomas on biopsy of these lesions. This finding has already been reported in earlier literature.[4] It

has been hypothesized that the pits are a result of the premature desquamation of most of the horny layer.

Other findings in Gorlin's syndrome are mesentric, ovarian and mammary cysts, uterine fibromas, renal calculi and calcification of the falx cerebri, falx cerebelli and dura or basal ganglia. Some patients also show a hyporesponsiveness to parathormone, which suggests a relationship to pseudohypoparathyroidism. Mental deficiencies and neurological disturbances have also been reported. Our cases had none of these findings.

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