PATHOLOGICAL DIAGNOSIS

Gorlin's Syndrome-A Case Report

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Abstract

Gorlin's syndrome is a rare disorder transmitted as an autosomal dominant trait. Most common phenotypic expression of this syndrome is a basal cell carcinoma (BCC). It is characterized by multiple skin lesions on head and neck region. We present a case of 49 year old male who presented with basal cell carcinoma at multiple sites simultaneously.

Key Words

Basal cell, Nevus, Syndrome, Carcinoma.

Introduction

Gorlin's syndrome also known as basal cell nevus syndrome (BCNS) and nevoid basal carcinoma syndrome, described by Clendenning (1) *et. al.* 1964, Gorlin (2) *et. al.* 1965, and Culter (3) *et. al.* 1979, is a genetically determined disorder, characterized by onset of multiple BCC. Its mode of inheritance is autosomal dominant with variable expressivity but about 60% patients have no family history. The tumour has varied clinical appearance and often look more like ordinary nevi, seborrheic keratosis or non descript papules (4). Microscopically, however, the picture is that of typical BCC (5).

Case Report

A 49 year old male patient presented with multiple papulo-nodular lesions over face, scalp and temporal region of 3 years duration (Fig-1). These lesions ranged from 0.3 cm to 1.0 cm in size, were soft, mobile and gradual in progression. Their appearance was variegated with some of them ulcerated. No regional lymph node was involved. There was no evidence of local bone involvement and distant metastasis. One of the lesions excised few months back had not shown recurrence so far. There was no history of spontaneous regression of any of the lesions. Patient had been operated for a mandibular cyst few years back. Two of the lesions, one from forehead and other from right cheek were excised and examined histopathologically. Microscopically both biopsies showed undifferentiated solid appearance of classical basal cell carcinoma (Fig-2).

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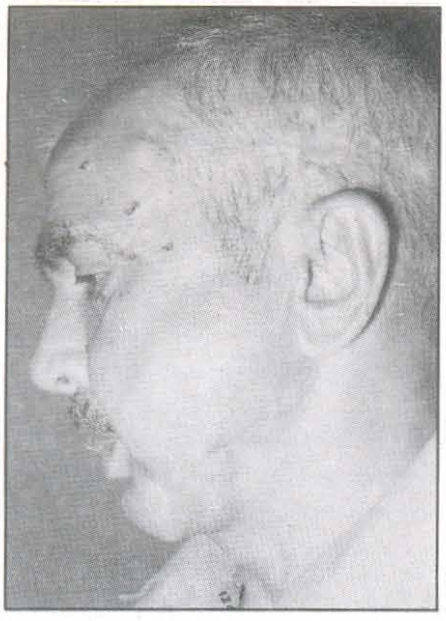

Fig. 1. Basal cell nevus syndrome. Two of these papules proved to be Basal cell carcinoma.

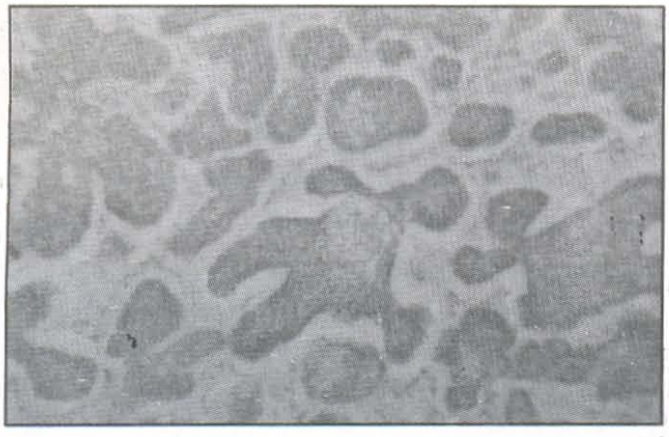

Fig. 2. Micrograph of basal cell carcinoma excised from cheek. (x400 X).

Discussion

BCC develops more frequently in several inheritable syndromes like Rasmussen syndrome, Rombo syndrome, Albinism, Xeroderma pigmentosa including Basal cell syndrome (4,6,7). It is also observed in linear unilateral basal cell nevus syndromes (5). The BCNS is a very rare disorder, usually occurs over forty years of age in 90% cases and is characterized primarily by 5 major findings (2) :–

1 Multiple basal cell carcinoma (BCC).

2 Pits of palms and sole.

3 Skeletal abnormalities especially of ribs.

4 Jaw cysts.

5. Calcification of falx.

In addition, tumours like medulloblastoma, fetal rhabdomyoma, cardiac fibroma and ovarian fibrosarcoma have been reported. Odontogenic keratocysts are characteristic and vertebral anomalies may be seen. Mental retardation, ocular and urogential abnormalities have also been described (8).

Except for multiple BCC, the incidence of other physical findings is highly variable and low. BCC are the most significant phenotypic expression of this disease. The life time incidence is more than 50% but total number and behaviour of diseases is quite heterogenous. These tumours may arise before adolescence but are not associated with aggressive behaviour (4).

The lesions of BCNS occur mostly on face and histologically all microscopic variants of ordinary BCC are seen (5).

While it is known that the syndrome is familial, exact pathogenesis is not understood. Ocurrence on head and neck suggests ultravoilet exposure as one of the factors. X-irradiation of such patients leads to striking increase in number of BCC within the treated field suggesting increased mutagenic potential. Chromosomal instability of DNA repair may be an inherent component of the syndrome.

The BCC originates from inter-follicular pluripotent stem cells differentiating towards follicular structures. The relation between BCC and pilosebaceous follicle is well established. Biochemical analysis of the keratin of BCC reveals that these tumours usually contain peptides that are characteristic of follicular epithelium. Some endo or exogenous component of sebum could contribute to tumour induction or promotion. Many cell surface markers are absent in BCC as compared to normal skin. Ricinus communis agglutinin 1 (RCA-1), β_2 microglobulin and C-3 binding properties associated with intermediate sized filaments are absent from BCC (9).

Amplification of C-myc oncogene and mutations involving P^{53} have been described. Although BCC can cause death (due to brain or other major organ invasion), it rarely produces metastasis (0.1%). Spontaneous regression is seen in 20 % cases (10).

Management demands close surveillance. Patients should be instructed to avoid sun exposure and use protective clothing and sunscreens. Excision is preferred treatment modality. Systemic retinoids have shown to cause decrease in BCC and rate of reappearance of new lesions but baseline activity quickly returns after discontinuation of therapy. These produce terminal differentiation in BCC. They increase tumouricidal activity of macrophages (4).

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