

**GORLIN'S SYNDROME: ATYPICAL CASE REPORT**

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None

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**Abstract**

Gorlin syndrome or basal cell nevus syndrome (BCNS) is a rare autosomal dominant disorder. The condition appears to have complete penetrance and variable expressivity, which makes clinical presentation among families variable. All known BCNS carry mutations in PATCHED gene. A 65 years old male patient presented with complaints of characteristic skin lesions on his face, back, palms since early adulthood. The lesions were pigmented nodules with characteristic border. The histopathology showed characteristic features suggestive of Basal Cell Carcinoma (BCC). This case was atypical due to appearance of lesions quite later in life.

**Key words:** Gorlin's Syndrome; Basal Cell Carcinoma; skin carcinoma

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**Introduction**

The name 'Gorlin' is associated with many genodermatosis like 'Gorlin Sign' in Ehler Danlos syndrome and Goltz-Gorlin syndrome of focal dermal hypoplasia and Gorlin syndrome. Gorlin Syndrome is also known as Basal Cell Nevus Syndrome (BCNS) or Nevroid Basal Cell Carcinoma Syndrome (NBCCS). It is an autosomal dominant genodermatosis characterised mainly by the presence of multiple basal cell carcinoma (BCC), jaw cysts and palmoplantar pits [1-3]. We would like to report an atypical case of Gorlin syndrome presented late in life.

**Case Report**

A 65 years old male patient presented with complaints of skin lesions on his face, back, palms since early adulthood. These lesions were insidious in onset and gradually increased in size. On dermatological examination six well defined, pigmented, non-tender nodules with raised and pearly border were seen on face (Fig. 1). A single ulcer of 2 x 3 cm with erythematous base and rolled borders was seen on right temporal region (Fig. 2). Multiple pits were present on both palms (Fig. 3). His hairs, nails and mucosa were normal. X-ray chest revealed bifid cervical rib and x-ray skull revealed falx cerebri calcification (Figs 4A and B). Dental examination was normal. On physical examination frontal bossing, kyphosis was present. Ophthalmic examination was normal. Other systemic examination were within normal limits. Routine laboratory investigations were within normal limits. With the characteristic morphology and other associated

features, a clinical diagnosis of BCC was made. The biopsy from lesion on back was consistent with BCC.

This case was atypical due to appearance of multiple basal cell carcinoma quite later in life as compared to general appearance of lesions since early childhood or birth.



**Figure 1.** Well defined, pigmented, non-tender nodules with raised and pearly border were seen on face.



Figure 2. Single ulcer of 2 x 3 cm with erythematous base and rolled borders was seen on right temporal region.



Figure 3. Multiple pits over both palms.

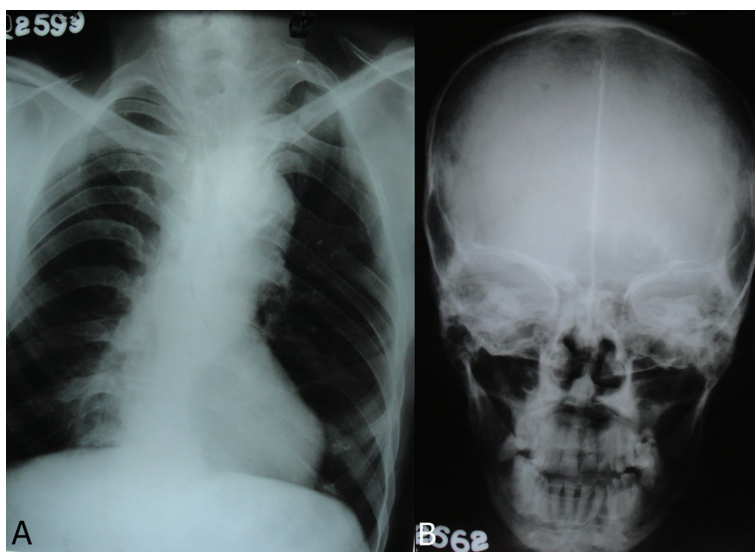


Figure 4 A and B. X-ray chest revealed bifid cervical rib and x-ray skull revealed falx cerebri calcification.

## Discussion

Gorlin syndrome or basal cell nevus syndrome (BCNS) is a rare autosomal dominant disorder. Prevalence of BCNS is estimated to be 1 in 60,000 to 1 in 1,20,000 [4]. In different studies, this condition appears to have complete penetrance and variable expressivity, which makes the clinical presentation among families variable. All known BCNS carry mutations in PATCHED gene. The diagnostic criteria for BCNS was put forth by Evans and Colleagues and modified by Kimoni in 1997 [4,5,7]. Accordingly, diagnosis of Gorlins syndrome could be established when two major or one major with two minor criteria are present as described below.

### Major criteria:

- 1) BCC before age of 30 or more than 2 BCC.
- 2) Odontogenic keratocyst before 15 years of age.
- 3) 3 or more palmar or plantar pits.
- 4) Falx cerebri calcification.
- 5) Rib anomaly.
- 6) First degree relative affected.
- 7) PTC gene mutation.

### Minor criteria:

- 1) Macrocephaly.
- 2) Congenital malformations - cleft lip, cleft palate, frontal bossing, hypertelorism.
- 3) Skeletal deformity – kyphosis, scoliosis.
- 4) Radiological abnormality – bridging of sella tursica, rib anomaly, Hemivertebra.
- 5) Ovarian fibroma.
- 6) Medulloblastoma.

Our patient has four major with two minor criteria which is diagnostic of Gorlin's Syndrome.

The management includes multidisciplinary approach. If the lesions are limited then surgery is indicated. Other treatment modalities include topical imiquimod 5%, laser ablation and strict avoidance of sun exposure. Oral isotretinoin at doses of 0.5-1mg/kg/day may cause regression of lesions of less than 1cm and may prevent new lesions [8]. High doses of retinoids were not effective and were associated with toxicity [4].

Early diagnosis and treatment is important to prevent long term complications of this syndrome that include malignancy, oromaxillofacial deformation and destruction. Aggressive BCC causes death of the patient as a result of tumour invasion to brain or other vital structures and medulloblastoma associated with syndrome causes death during infancy [6]. Genetic counselling also plays a vital role.

This case is atypical due to appearance of lesions quite later in life.

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