Naevoid basal cell carcinoma syndrome

Sir,

Nevoid basal cell carcinoma syndrome (NBCCS), also known as Gorlin syndrome, is a hereditary condition characterized by a wide range of developmental abnormalities and a predisposition to neoplasms. The syndrome, delineated by Gorlin and Goltz in 1960, was first reported in 1894 and has been given several different names including Gorlin syndrome, Gorlin-Goltz syndrome, Nevoid basal cell carcinoma syndrome (NBCCS), multiple nevoid basal-cell carcinoma syndrome (MNBCCS), multiple basal-cell carcinoma syndrome, basal cell nevus syndrome (BCNS), multiple basalioma syndrome, fifth phacomatosis, jaw cysts-basal cell tumors-skeletal anomalies syndrome, odontogenic keratocytosis-skeletal anomalies syndrome, multiple nevoid basal-cell epithelioma-jaw cysts-bifid rib syndrome, hereditary cutaneomandibular polyoncosis and finally epitheliomatose multiple generalisee.[1]

The estimated prevalence varies from 1/57,000 to 1/256,000, with a male-to-female ratio of 1:1.[2] Main clinical manifestations include multiple basal cell carcinomas (BCCs), odontogenic keratocysts of the jaws, hyperkeratosis of palms and soles, skeletal abnormalities, intracranial ectopic calcifications, and facial dysmorphism (macrocephaly, cleft lip/palate and severe eye anomalies).

In the current case series, we present three confirmed

Table 1: Characteristics of the three patients with their manifestations

Sex	Age (yrs old)	Characteristics
F	18	Multiple BCC† [Figures 1 and 2], Meduloblastoma‡, Seizure, Palmar pits† [Figure 3], a broad nasal bridge with ocular hypertelorism and frontal bossing‡, Skull punch out lesions
F	35	Multiple BCC†, Odontogenic mandibular keratocyst†
M	45	Multiple BCC†, Congenital Kyphoscoliosis‡ [Figure 4], Frontal bossing‡

†Major criteria; ‡Minor criteria

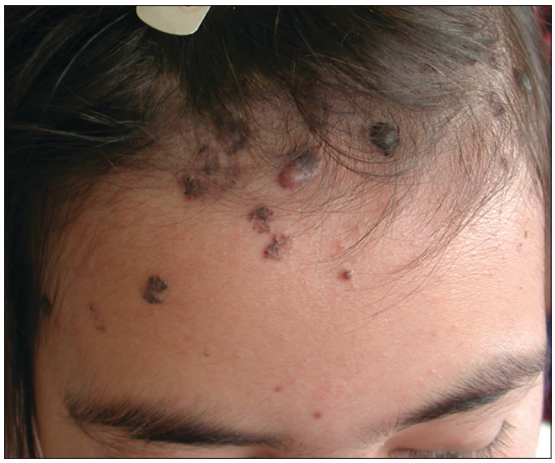

Figure 1: Multiple BCCs on the scalp

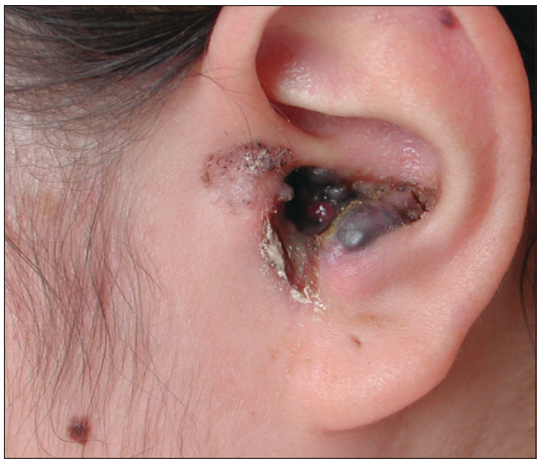

Figure 2: Large BCC on the ear

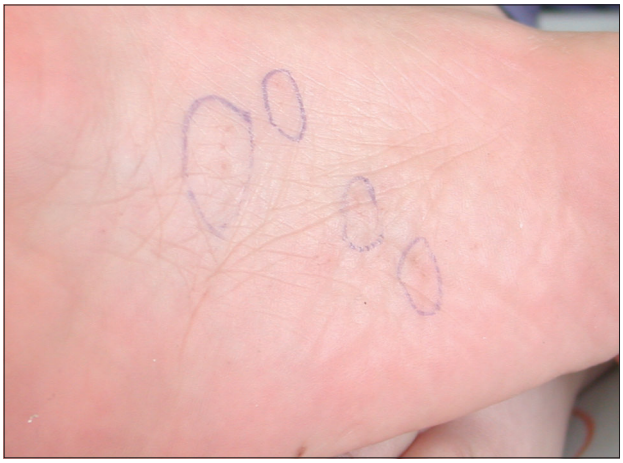

Figure 3: Palmar pits

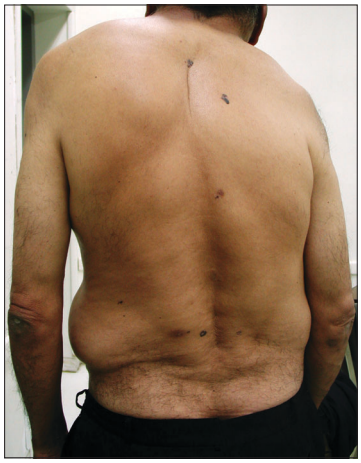

Figure 4: Kyphoscoliosis along with BCC on the back

cases of NBCCS with review of their manifestations [Table 1].

The diagnostic criteria for NBCC was put forth by Evans and Colleagues and modified by Kimoni in 1997. According to him, diagnosis of Gorlin syndrome can be established when two major or one major and two minor criteria as described below, are present.

Major criteria include more than two basal cell carcinoma or one basal carcinoma at younger than 30 years of age or more than10 basal cell nevi, any odontogenic keratocyst (proven on histology) or polyostotic bone cyst, three or more palmar or plantar pits, ectopic calcification lamellar or early at younger than 20 years of age, falx cerebri calcification and positive family history of nevoid basal cell carcinoma.

Minor criteria include congenital skeletal anomaly, bifid, fused, splayed, missing or bified rib, wedged or fused vertebra, occipital-frontal circumference more than 97 percentile with frontal bossing, cardiac or ovarian fibroma, medulloblastoma, lymphomesentric cysts, and congenital malformations such as cleft lip or palate, polydactylism or eye anomaly (cataract, coloboma, microphthalmus).[3,4]

In our first case, two major (multiple BCCs at younger than 30 years old, and palmar pits) and one minor (medulloblastoma) criteria were met, which were indicative of Gorlin syndrome. The second case had two major criteria (multiple BCCs before 30 years and odontogenic keratocyst), which confirmed Gorlin syndrome. The third case had one major criterion (multiple BCCs) and two minor criteria (congenital skeletal anomaly and frontal bossing) which confirmed Gorlin syndrome.

Early diagnosis and treatment is important to prevent long term squeals of this syndrome that include

malignancy and oromaxillofacial deformation and destruction. Aggressive BCC causes death of the patient as a result of tumor invasion to the brain or other vital structures and medulloblastoma associated with the syndrome causes death during infancy.[5] Because of recurrence of odontogenic keratocysts, varying degrees of jaw deformity may result from operations for multiple cysts.

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DOI: 10.4103/0378-6323.58690 - **PMID:** 20061742

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