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# Nevoid Basal Cell Carcinoma Syndrome: A case report

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***Abstract:*** *The Nevoid Basal Cell Carcinoma Syndrome (NBCCS) also known as Gorlin-Goltz Syndrome is an Autosomal Dominant Disorder characterized bymultiple Odontogenic Keratocysts (OKCs) in the jaw, multiple basal cell carcinomas (BCCs) and skeletal abnormities. This paper reports a case of a 20-year old male patient with NBCCS emphasizing on its clinical and radiographic manifestations along witha brief literature review.*

***Keywords:*** *NBCCS, basal cell carcinoma, multiple keratocystic odontogenic tumors, Gorlin- Goltz syndrome*

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## I. Introduction

The Nevoid Basal Cell Carcinoma Syndrome(NBCCS) was first reported by Jarisch and White 1894.[1,2] It was only in 1960 that Robert James Gorlin and William Goltz described the classical triad of multiple Basal Cell Carcinomas (BCCs), Odontogenic Keratocysts (OKCs) and Bifid Rib.[3]NBCCS has an autosomal dominant pattern of inheritance with a prevalence of about 1 per 60,000.[4]

The peak incidence of OKCs not associated with NBCCS is 20-30 years.However, in the NBCCS, OKCs occurs in much younger individuals. Males have a slightly higher predilection for OKCs not associated with NBCCS (1:0.62) whereas OKCs in NBCCS have a higher predilection in females (1:1.22).[5]OKCs associated with NBCCS are more commonly seen in the mandibular molar-ramus region.[6]Reporting herewith, a patient with classical signs of this syndrome.

## II. Case Report

A 20-year old male patient reported to the department in the year 2018with the complaint of missing teeth. He gave a history of pus discharge from his gums 3 years back due to which multiple teeth were extracted.He also reported that a surgical procedure was done in the mandibular anterior region along with the extractions. His past medical history and family history werenon-contributory. On extraoral examination there were two melanotic papules notedon the left side of his face. One papule roughly ovoid in shape in the right infraorbital regionmeasured approximately 3x2 mm in size (N1) and another circular in shape in the left nasolabial fold measured approximately 2 mm in diameter (N2) (Figure 1). On general examination, multiple palmar pits were seen on the right palm. His right hand showed the presence of an accessory finger. Intraorally, 17, 23, 24, 25, 33, 32, 31, 41, 42, 43, 45, 46were clinically missingand showed no evidence of swelling or expansion.There was also no evidence of pus discharge from the gingiva. Resorption of the alveolar bone was noted in the mandibular anterior region. On palpation,alveolar bone was hard in consistency.According to the patients' history and clinical examination, presence of a cystic lesion was suspected in the mandibular anterior region. Hence, a provisional diagnosis ofNBCCS was given.

The case was further investigated with radiographic and histopathological analysis. An orthopantomograph was then taken which revealed multiple unilocular radiolucencies in the maxilla and mandible. Loss of coronal tooth structure was seen with respect to tooth 36 with a uniform radiolucency around mesial and distal rootsand ill-defined non-corticated margins. (Figure 2). The radiolucencies in the maxilla were well appreciated on a CBCT, one of which extended from 16 to 21 with ill-defined periphery and uniform radiolucency within. Another ovoid radiolucency was seen in 24, 25 region with well defined corticated periphery and uniform radiolucency within. The mandible showed presence of twosimilar cystic lesionsin 38 and 45 regions respectively. An ill-defined radiolucent area was noted in the anterior mandibular region with sparse bony trabeculae. These lesions showed no bucco-lingual cortical bone expansion.A radiographic diagnosis of multiple odontogenic cysts and a differential diagnosis of Odontogenic Keratocyst in relation to16 to 21, Residual Cysts in relation to 24 to 25, 38, 33 to 43, 45and Radicular Cyst in relation to 36was given.

Skull and chest radiographs were taken toevaluate the possibility of the presence of other findings associated with multiple cystic lesions. Lateral cephalogram showed bridging of sella turcica. PA skull revealed calcification of falx cerebri. Presence of a bifid $4^{th}$ rib on the right side was revealed on a chest radiograph(Figure 3 A, B, C).

An incisional biopsy of the lesion i.r.t. 18 to 21was taken along with cystic aspirate which revealed yellowish-white cheesy material. The protein content of the aspirate was found to be 4 mg/dl. The histopathological report of the lesion i.r.t. 16 to 21 confirmed the diagnosis of OKCs.

An excisional biopsy of N1 and N2 nevi was performed which revealedAdenoid Basal Cell Carcinoma in N1 and Pigmented Basal cell Carcinoma in N2. The above clinico-radiological and histopathological findingsaffirmed thediagnosis of NBCCS.

The cysts were then enucleated and Carnoy's solution was used to carry out chemical cauterization under general anaesthesia in Oral and Maxillofacial Surgery Department. Histopathological evaluation of enucleated cysts confirmed the diagnosis of Odontogenic Keratocysts and Radicular Cyst in relation to 36. The patient was recalled after a month, 3 months and now being evaluated every 6 months in our department.

## III. Discussion

NBCCS is characterised byOKCs, BCCs with its manifestations also affecting skeletal, ophthalmic, dermatological, neurological systems.It is mainly caused by mutations in the patched tumor suppressor gene 1 (PTCH 1), on chromosome 9q22.3.[7] Recent genetic studies also suggest involvement of PTCH 2 and SUFU markers in this syndrome.[8]

Since NBCCS is aninheritable condition,offspring of affected individual has 50% chance of developing this syndrome.[9]Hence genetic screening and counselling of patients and family members plays an importantrole in the early diagnosis and management of suspected disease.

Diagnostic criteria for NBCCS was first given by Evans *et al.* (1991)[10]and by Kimonis *et al.* (1997).[11]Diagnosis of NBCCS requires the presence of 2 major criteria or 1 major and 2 minor criteria.[11] The present case fulfils5 major criteria (*) and 2 minor criteria (#).(Refer Table 1)

As reported in literature, the incidence of OKCs ranges from 62% to 100% in patients with NBCCS.[12,13,14,15]. Clinically, the lesions tend to recur and grow aggressively after surgical treatment. It is believed that higher recurrence of OKC is associated with the presence of daughter cysts or epithelial islands in the cyst wall. Such was demonstrated in a report by Myoung et al.[16] The mandible is more often involved than the maxillawith the posterior regions being most commonly affected.In the present case, 3of 5 cysts were located in the mandible and 4 of 5 were located in the posterior region.

The conservative methods of treating OKCs includesimple enucleation with or without curettage and marsupialization whereasperipheral ostectomy, chemical curettage with Carnoy's solution and resection are more aggressive methods.After reviewing literatures regarding the treatment and prognosis of OKCs,Blanas et al stated that "OKC hasa 17–56% recurrence rate if simple enucleation is carried out in isolation, and if adjunctive treatment is added, such as the application of Carnoy's solution or decompression before enucleation, the recurrence rate is reported to be between 1–8.7%".[17]

Long follow-up periods along with careful monitoring of patients by radiographic imaging should be done annually in order to minimize abnormalities post treatment. Patients should be counselled to prevent ionizing radiations and harmful UV exposure that increase the risk of developing BCC.

## IV. Conclusion

In the present case, the patient remained undiagnosed until 20 years of age. It was only when the patient seeked dental care for his missing teeth, the diagnosis of NBCCS was made. It is of utmost importance for early diagnosis of NBCCSsincecomplications such as malignant tumors of the brain and skin can be managed early and deformities related to the jaw cysts can be prevented.This case reinforces the idea that the dentist has animportant role in prompt diagnosis and referraltospecialists for evaluation of other systems.

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**Figure 1:** Extraoral examination revealing 2 melanotic papules on the left side of the face marked N1 and N2 (arrow).

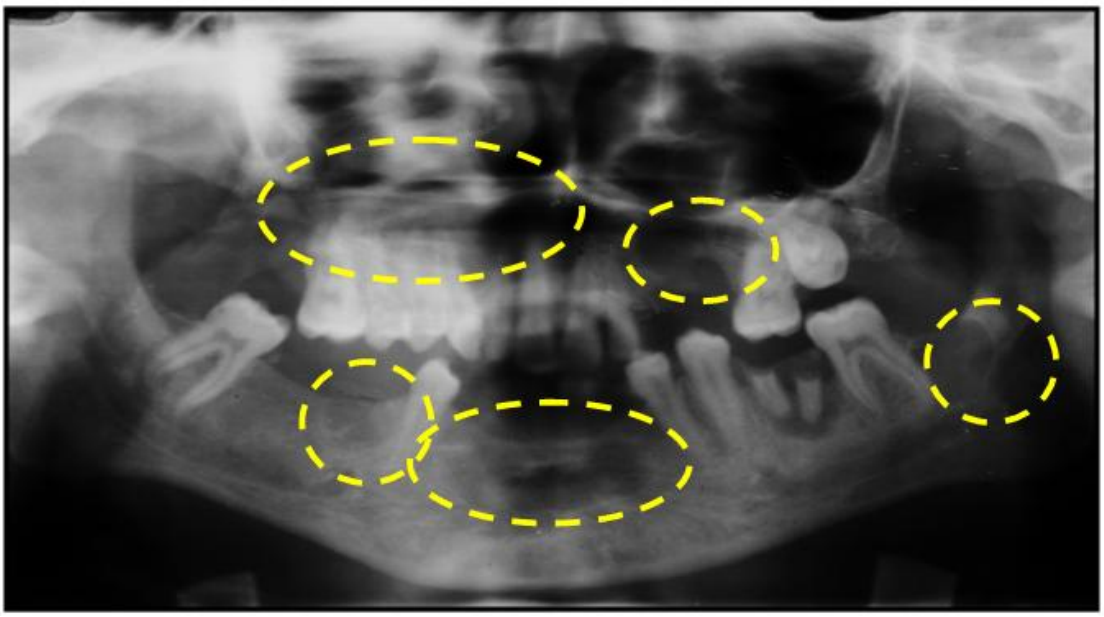

**Figure 2:** A screening orthopantomograph revealing multiple unilocular radiolucencies in the maxilla and mandible (circles).

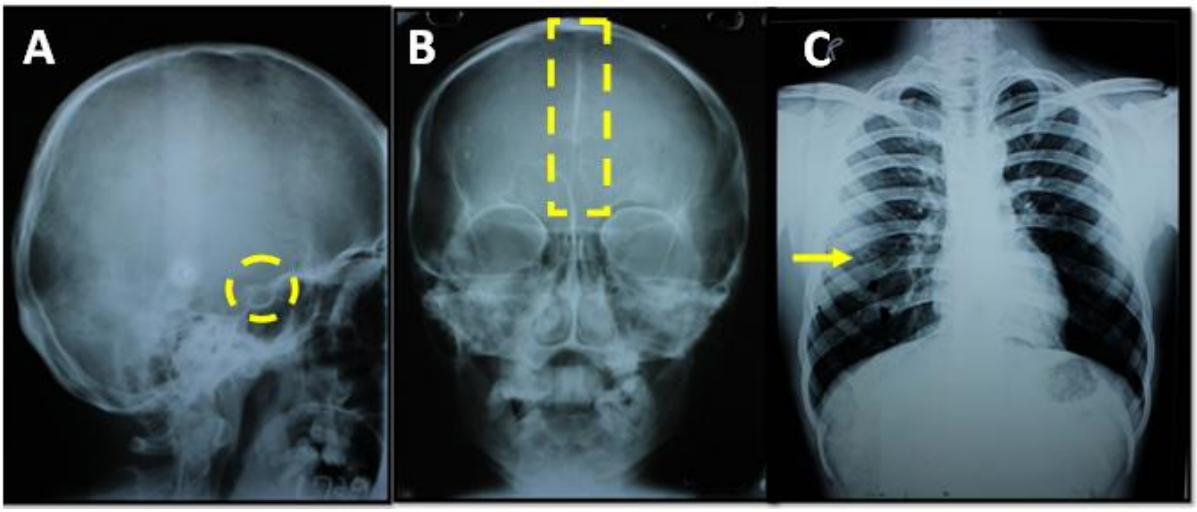


**Figure 3:** A. Lateral cephalogram showing bridging of sella turcica (circle), B. PA skull showing calcification of falx cerebri (box) and C. Chest radiograph showing bifid 4th rib on the right side (arrow).

**Table 1:** Major and minor criteria noted in present case

| "Criteria given by Veenstra-Knol H. E. et al."[18] | Features noted in present case |
|---|---|
| **MAJOR CRITERIA** | |
| Multiple (>2) basal cell carcinomas, or one diagnosed under 20 years | ✓* |
| Odontogenic keratocysts proven by histology | ✓* |
| ≥3 palmar or plantar pits | ✓* |
| Ectopic calcification (lamellar or early falx) | ✓* |
| Bifid, fused or markedly splayed ribs | ✓* |
| Family history of NBCCS | |
| **MINOR CRITERIA** | |
| Macrocephaly determined after adjustment for height (≥97th percentile) | |
| Bridging of the sella turcica | ✓# |
| Vertebral anomalies such as hemivertebrae | |
| Fusion or elongation of the vertebral bodies | |
| Defects of the hands and feet | |
| Cleft lip or palate | |
| Polydactyly | ✓# |
| Cardiac or ovarian fibroma | |
| Medulloblastoma | |
| Other skeletal abnormalities such as Sprengel deformity, marked pectus deformity, marked syndactyly of the digits | |
| Eye anomaly: cataract, coloboma, microphthalmia | |
| PTCH mutation | |

** major criteria # minor criteria*