

# The Role of Collaborative Diagnosis in Identifying Nevroid Basal Cell Carcinoma Syndrome: A Vigilant Radiologist Helping a Surgeon

Chakraborty P<sup>1\*</sup>, Kumari R<sup>1</sup>, Singh AP<sup>2</sup> and Jain RK<sup>3</sup>

<sup>1</sup>Department of Otorhinolaryngology, Banaras Hindu University, Varanasi, India

<sup>2</sup>Department of Radiodiagnosis and Imaging, Banaras Hindu University, Varanasi, India

<sup>3</sup>Professor and Head, Department of Otorhinolaryngology, Banaras Hindu University, Varanasi, India

**\*Corresponding author:** Priyanko Chakraborty, Department of Otorhinolaryngology, Institute of Medical Sciences, Banaras Hindu University, Varanasi, India, E-mail: cpriyanko@gmail.com

## Case Report

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## Abstract

Nevroid basal cell carcinoma syndrome is a rare inherited condition caused by mutations in the PTCH1 gene. It causes a wide range of developmental abnormalities. These patients are encountered by otolaryngologists and radiologists initially presenting with the odontogenic keratocysts affecting maxilla and mandible. Multiple basal cell carcinomas, pits in palmoplantar region and jaw cysts are cardinal features. Often surgeons are unfamiliar with the different manifestations leading to failure to diagnose the condition. A vigilant radiologist can help in the diagnosis and potentially preventing the sequelae.

**Keywords:** Odontogenic keratocysts; Nevroid basal cell carcinoma; Bifid ribs; Gorlin goltz syndrome

## Case Report

A 27 year old male, presented in the department of otolaryngology with progressive bilateral maxillary swelling since childhood causing substantial disfigurement, there was history of intermittent nasal obstruction; with no history pain, sinus, facial heaviness or epiphora. The patient had no other complaints. The patient was provisionally diagnosed as a case of odontogenic tumor and a contrast enhanced CT scan of face, nose and paranasal sinuses, 1-1.5 mm sections, both in soft tissue and bone window settings; axial, coronal and sagittal view was advised. In the department of radiology, contrast enhanced CT scan of face and paranasal sinuses (bone and soft tissue window) showed multiple cystic expansile lesions with unerupted teeth suggestive of odontogenic keratocysts. Incidentally the visualized portion of the brain and cranium in the CT scan of face

revealed calcification of falx cerebri and tentorium cerebelli raising the suspicion of nevroid basal cell carcinoma syndrome. The patient was then thoroughly investigated by the radiologists and a chest X-Ray was done which showed the classical picture of bifid and fused ribs. And finally the case was radiologically reported as a possible case of nevroid basal cell carcinoma syndrome. This vigilant reporting encouraged the otolaryngologists to closely examine the patient for any basal cell nevus or carcinoma. An asymptomatic retro-auricular ulceronodular lesion was detected which was present there for over an year. It looked suspicious and was excised with adequate margins (1 cm from the palpable margins) and on histopathological examination showed features of basal cell carcinoma. The odontogenic keratocyst was excised too with a guarded prognostication for relapse. Also multiple nevi were

present at the back. No family history of facial swellings and basal cell naevi was present (Figure 1-3).

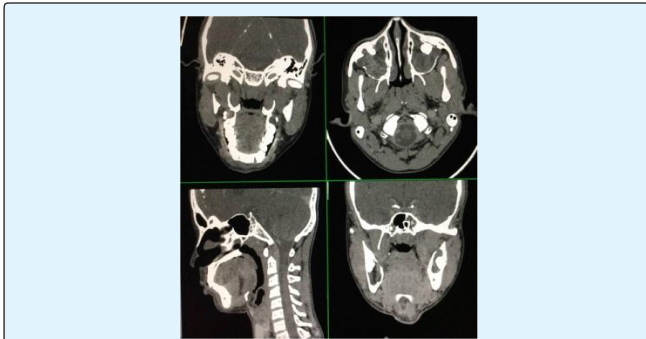


Figure 1: Coronal, sagittal and axial scans of CT scan face including cranium showing multiple odontogenic keratocysts of maxilla and calcification of falx cerebri.



Figure 2: Chest X-Ray PA view showing multiple bifid and fused ribs.



Figure 3: Clinical photograph showing post aural basal cell carcinoma.

## Discussion

Nevoid basal cell carcinoma syndrome was first reported by Jarish in 1894 [1]. Gorlin and Goltz in 1960 gave the complete description and of the new syndrome [1]. Nevoid basal cell carcinoma usually present at the first, second or third decade [2]. There are so many other confusing names of the syndrome including Gorlin syndrome, basal cell nevus syndrome, syndrome of jaw cysts, hereditary cutaneous mandibular polyonocosis or jaw

cyst-basal cell nevus-bifid rib syndrome [3,4]. Gorlin had suggested the term nevoid basal cell carcinoma syndrome, however all the affected patients did not have basal cell carcinomas. Nevoid basal cell carcinoma is inherited by autosomal dominant mode, but can have a variable phenotypic penetration, or can have sporadic presentation as seen in our case [5]. This syndrome is associated with a wide spectrum of developmental anomalies and neoplasms. Keratocystic odontogenic tumor or odontogenic keratocyst is usually the first feature of the syndrome. The tumor is typically found as incidental radiographic findings or may present as jaw swellings. It has been suggested that multiple OKCs alone may be confirmatory of the syndrome [6]. Although benign, the recurrence rate after excision of OKC is very high, ranging from 12 to 62.5% and multiple recurrences are not unusual [7]. The PTCH 1 gene, the human homologue of the *Drosophila* segment polarity gene, has been seen to be involved in the development of Gorlin-goltz syndrome [8,9].

Odontogenic keratocystic tumors in Gorlin-Goltz syndrome comprise unilocular or multilocular radiolucencies of the mandibular body, angle, or ramus [2]. In children and adolescents, the cysts may cause displacement of developing teeth and delayed dental development with associated impacted tooth [7]. The most frequent skin lesions of Gorlin-Goltz syndrome are cutaneous basal cell carcinomas, benign dermal cysts, and palmar and plantar keratosis or pits [2,7]. Ectopic calcification of falx cerebri, tentorium cerebella, and bridged sella may also be detected radiologically [7]. The patient usually has cranio-facial anomalies comprising of frontal and parietal bossing and broad nasal root which may be associated with ocular hypertelorism [2]. Thoracic cage anomalies such as bifid and fused ribs may be present as classically seen in two of our cases. Syndactyly or polydactyly of toes may be the other skeletal abnormalities along with kyphosis and scoliosis [2]. The diagnosis of Gorlin-Goltz syndrome requires the presence of two major or one major and two minor criteria [7,9] (Table 1).

Major Criteria	Minor Criteria
Multiple Bcc	Macrocephaly
Odontogenic Keratocyst	Congenital Malformation
Palmar/ Plantar Pits	Skeletal Anomaly
Calcified Falx Cerebri	Radiologic Anomaly
Bifid/ Fused Rib	Ovarian Fibroma
1 <sup>st</sup> Degree Relative	Medulloblastoma

Table 1: Criteria for Diagnosis. Two major or one major and two minor criteria should be present [7,9].

The clinical management includes surgical treatment (enucleation and peripheral ostectomy) of the lesions [10]. Addressing the risks associated with syndrome, the development of neoplasm's on sun or radiation exposure, recurrent odontogenic keratocysts etc., warrants utmost importance. Thus a regular follow up, interdisciplinary cooperation for the diagnosis, treatment and rehabilitation is required. All the patients were counseled regarding the syndrome and are kept under active follow-up/surveillance for early detection of cutaneous malignancies and other sequelae of the syndrome.

### Conclusion

The patients affected by Gorlin-Goltz syndrome must be evaluated by several relevant specialists to precisely confirm the diagnosis. Early diagnosis and treatment may reduce the severity of the long term sequelae of Gorlin-Goltz syndrome including malignancy and oral maxillofacial deformation and destruction.

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