CASE REPORT

# Gorlin-Goltz Syndrome

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## Abstract

A 12 years old girl was presented with bilateral swellings on angle and body of mandible. On general physical examination, there were polydactyly and papular lesions on arm. Histopathology of mandibular lesions revealed odontogenic keratocysts. Marsupialization of the cysts followed by enucleation was done. The patient was reviewed every six months and there was no recurrence at the end of two years.

**Key Words:** *Odontogenic keratocyst. Marsupialization. Enucleation. Gorlin-Goltz syndrome. Nevoid basal cell carcinoma syndrome.*

## INTRODUCTION

Nevoid Basal Cell Carcinoma Syndrome (NBCCS), also known as Gorlin-Goltz syndrome, is an autosomal dominant disorder characterized by a predisposition to neoplasms and other developmental abnormalities. Gorlin and Goltz described the classical triad of multiple basal cell carcinomas, odontogenic keratocysts in the jaws and bifid ribs that characterize the diagnosis of this syndrome.[1] In addition to this triad, calcification of the falx cerebri, palmar and plantar epidermal pits, spine and rib anomalies, facial milia, ocular malformation, medulloblastomas, cleft lip or palate, and other developmental anomalies have also been described as features of the syndrome. The prevalence of NBCCS has been estimated to be about 1 per 60,000. This syndrome probably presents itself in all ethnic groups.[2]

The tumor suppressor gene called PATCHED (PTCH 1), located in the 9q22. 3-3.1 chromosome has been identified as cause of NBCCS. However, mutations in others genes such as PTCH2, Smmothened (SMO) and Sonic HedgeHog (SHH) have also been reported in isolated cases of basal cell carcinoma and medulloblastoma.[3,4] The purpose of reporting of this case is to highlight the importance of early detection, treatment of morbidities and regular follow-up.

## CASE REPORT

A 12 years old girl reported to dental OPD with complaint of swelling on both side of angle and right body of mandible for the last two months. On general physical examination there were small papular skin coloured lesions on right arm of the patient and there was polydactyly. X-rays Orthopantomogram (OPG) of the face revealed multiple cystic lesions in mandible (Figure 1). X-rays of chest and spine revealed bifid ribs (Figure 2) and spina bifida. Pelvic examination revealed no such lesions. Histopathology of the cystic lesions of mandible revealed an odontogenic keratocyst of mandible. Based on these findings, the patient was diagnosed as a case of "Gorlin-Goltz syndrome". Patient's father was counseled and explained about the disease and its complications.

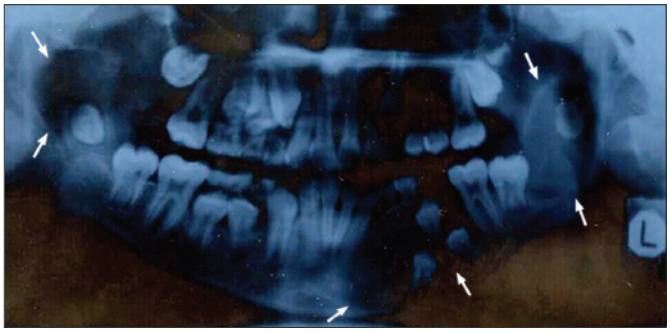

**Figure 1:** X-ray OPG: arrows showing multiple cystic lesions.

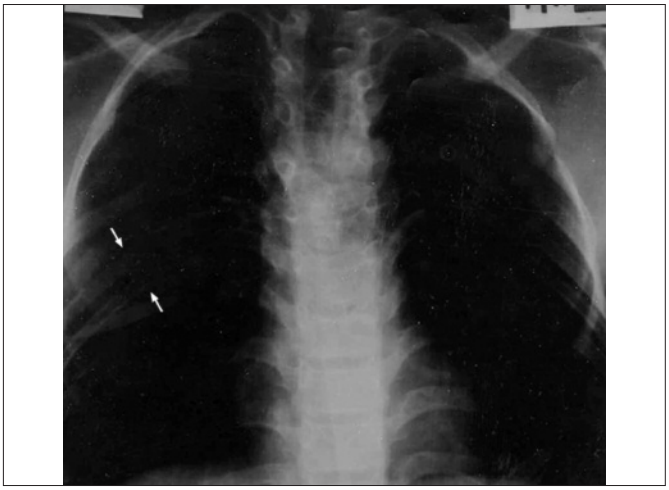

**Figure 2:** X-ray Chest (arrows) showing bifid ribs and upper dorsal spina bifida.

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*Received: December 31, 2012; Accepted: March 06, 2014.*

Treatment plan was outlined as marsupialization of cystic lesions and then enucleation to preserve the permanent dentition. Bilateral vestibular incision was given on both sides of angle of mandible, bone windows made, cystic fluid drained out and normal surrounding tissue tucked in the cavities and packed with Bismuth Iodoform Paraffin Paste (BIPP).

The patient was advised for a regular follow-up on weekly basis for the removal of old BIPP and flushing of the cavities with saline. After two months, size of the cavities was markedly reduced. Enucleation was done to remove the complete cystic lesions and wound was closed. Healing was excellent. The patient was reviewed every six months for two years. There was no recurrence at the end of this period.

## DISCUSSION

Several studies have described Odontogenic Keratocysts (OKCs), basal cell naevi and skeletal anomalies as the principal clinical features of NBCCS. However, according to Manfredi *et al*.,[5] the diagnostic criteria of NBCCS requires the presence of two major, or one major and two minor criteria. Major criteria include the presence of more than two basal cell carcinomas or one under the age of 20 years, histologically proven OKCs of the jaw, cutaneous palmar or plantar pits, and bifid ribs.[4] Minor criteria include orofacial congenital malformations, skeletal and radiological abnormalities, ovarian fibroma and medulloblastoma.[5] The present case showed multiple OKCs in the mandible, rib anomalies, spina bifida, skin lesions and polydactyly. This confirmed the diagnosis of NBCCS or Gorlin-Goltz syndrome.

According to Marotto *et al*., some of the most common clinical findings of the syndrome were discovered through radiography.[6] A panoramic radiograph showed radiolucent areas in the mandible, suggesting the presence of OKCs. Chest radiograph indicated the presence of rib anomalies, posterio-anterior view of the skull and computed tomography scan of the head and neck showed calcification of the cerebral falx and spina bifida, which according to Amezaga *et al*. are characteristic of the syndrome.[2] OKCs are among the most consistent and common features of NBCCS. These are found in 65 - 100% of affected individuals. The mandible is involved more frequently than the maxilla and the posterior regions are the most commonly affected sites.

There are two methods for the treatment of OKCs, a conservative and an aggressive. In the conservative method, simple enucleation with or without curettage and marsupialization are suggested.[6,8] Aggressive methods include peripheral ostectomy, chemical curettage with Carnoy's solution and resection.[8]

Radical interventions as enucleation with shaving of surrounding bone or sometimes resection might contribute to prevent recurrences and to improve the prognosis. In children who have not yet developed permanent dentition, conservative management should be considered first because an aggressive operation can have an adverse effect on teeth development, the eruption process and the development of the involved jaw.[9] Thus, younger patients usually receive more conservative than aggressive treatment. Although some authors believe that simple enucleation might be the most appropriate conservative method for the treatment of OKCs, others have shown the successful treatment of large or multiple OKCs using the marsupialization followed by enucleation as in this case. Furthermore, it has been reported that marsupialization followed by enucleation results in the lowest recurrence rate among those undergoing conservative treatment.[5,10] Histopathological examination of the removed tumors should be performed to provide definitive diagnosis. In this case, the microscopic analysis confirmed the diagnosis of OKCs indicating the need for monitoring of the disease. Long follow-up is suggested for this disease. In order to minimize the secondary morbidities after the treatment, patients with OKCs should be observed carefully by radiographic imaging particularly during the first year.[11] Moreover, early diagnosis is important for counselling of patients to prevent harmful exposure to ultraviolet and ionizing radiations that increase the risk of developing basal cell carcinoma. The patient in this case study is also under the care of dermatologist for regular follow-up for early diagnosis and treatment of BCCs.

Gorlin-Goltz syndrome is of particular interest to the oral and maxillofacial health experts. Proper evaluation and characterization of the clinical features is of utmost importance for the correct diagnosis and early treatment of affected patients. In order to establish early diagnosis of NBCCS, specialists should carry out clinical and imaging examinations in early ages of life.[6]

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