

Case Report

Gorlin-Goltz syndrome- Two case reports

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ARTICLE INFO



Keywords:

Gorlin-Goltz syndrome, Nevoid basal cell carcinoma, odontogenic keratocyst, Carnoy's solution

ABSTRACT

Gorlin-Goltz syndrome it is also referred to as Nevoid Basal cell carcinoma syndrome(NBCC) or Basal cell nevus syndrome is a rare autosomal dominant inherited condition involves multiple organ system. It consists of classic triad of basal cell carcinomas, multiple odontogenic keratocysts and skeletal deformities along with other defects. These multiple jaw cysts have more recurrence rate due to their aggressive behaviour and immediate treatment is mandatory. Immediate enucleation and curettage and follow up of patient is necessary.

Introduction

Gorlin-Goltz syndrome or Nevoid basal cell carcinoma syndrome is an infrequent multisystemic disease that is hereditary condition and it is transmitted as an autosomal dominant trait with high or almost 100% penetrance and varying expressivity.¹ The first case of skin manifestation of the nevoid basal-cell carcinoma syndrome or the Gorlin-Goltz syndrome was described by Jarisch and White in 1894.² In 1960, Gorlin. The minimum prevalence is one in every 57,000 although 1 in every 200 patients with basal-cell carcinoma present NBCC and the proportion is much larger with patients that develop basal-cellular carcinoma before the age of 19.² NBCCS characterized mainly by presence of multiple odontogenic keratocyst (75%), basal cell carcinoma(50-97%), bifid ribs(40%), palmar and planter

pits(60-90%) and ectopic calcification of the falx cerebri(37-79%).³

CASE REPORT-1:

A 13 year old male child reported to the department of oral medicine and radiology AME'S dental college and hospital Raichur, with the chief complaint of intraorally a firm painless mass on the right side of body of mandible. The duration of swelling was 8-9 months and growth was slow in onset with no discharge. On examination the swelling was firm, non-tender and extending from the region of 43 to 46(Fig-1). General examination revealed frontal bossing, hypertelorism and depressed nasal bridge and palmar pits(Fig-1). OPG(Fig-2) show ed multiple lytic lesions in the maxillary and mandibular jaws associated with unerupted permanent

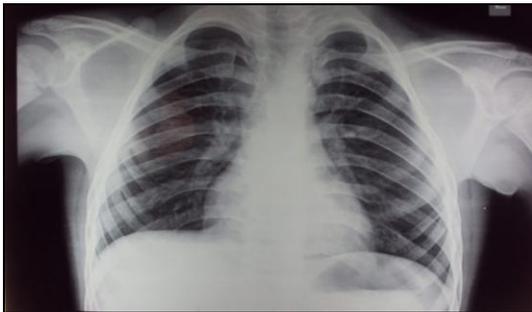
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(C-1) FIGURE-1- CLINICAL PHOTOGRAPHS



(C-1) FIGURE-2 - OPG PRE-OP

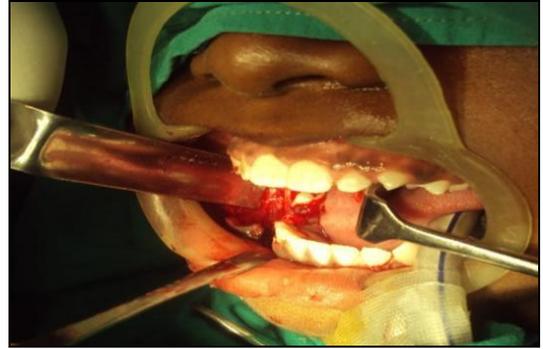


(C-1) FIGURE-3 . CHEST RADIOGRAPH

teeth displaced from their normal positions. The chest radiograph showed 3rd, 4th and 5th bifid ribs on right side and on left side 4th rib is bifid (Fig-3). The clinical and radiological findings were confirmatory for Gorlin-Goltz syndrome. Surgical enucleation of OKC followed by chemical cauterization with Carnoy's solution was planned for the patient and was followed up (Fig-4 & Fig-5) for the period of 3 yrs without any recurrence.

CASE REPORT-2:

A 28 year old male patient with complaint of pain and slight swelling on the right side of mandible since 6-8 months. Pain was sudden in onset, intermittent and throbbing in nature. He gives history of extraction of 12, 13 and 46, 2 years back. And he gives history of



(C-1) FIGURE-4. -SURGICAL PHOTOGRAPH.



(C-1) FIGURE-5. POST-OP. AFTER 3YR

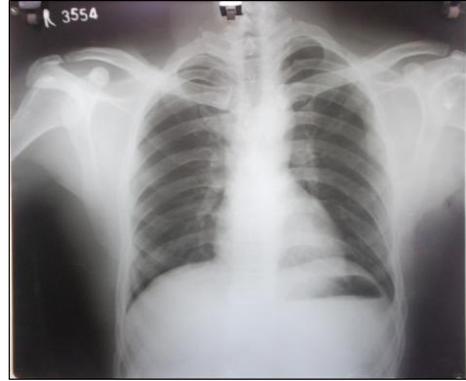
paraesthesia of lower lip region since 6 months (Fig-6). General examination revealed macrocephaly, frontal bossing, hypertelorism, prominent supraorbital ridges, depressed nasal bridge, mandibular prognathism and plantar pits (Fig-7). OPG revealed multiple lytic lesions in the right maxillary and in both right and left mandibular jaw (Fig-8). The chest radiograph showed bifid ribs bilaterally on 5th rib (Fig-9). The clinical and radiological findings were confirmatory for Gorlin-Goltz syndrome. Surgical enucleation of OKC followed by chemical cauterization with Carnoy's solution was planned for the patient (Fig-10) and was followed up for the period of 6 months.

DISCUSSION:

NBCCS was first described by Jarisch and White in 1894⁴ and later established as a unique syndrome by Gorlin and Goltz in 1960. The pathogenesis of NBCCS is attributed to abnormalities linked to the long arm of chromosome 9 (q22.3-q31) PCTH1 genes with no



(C-2) FIGURE-6-CLINICAL PHOTOGRAPH



(C-2) FIGURE-9.CHEST RADIOGRAPH.



(C-2) FIGURE-7.PLANTAR PITS.



(C-2) FIGURE-10.SURGICAL PHOTOGRAPH.



(C-2) FIGURE-8.OPG

apparent heterogeneity.⁵

The diagnosis of the Gorlin-Goltz syndrome is made clinically by using the criteria which are suggested by Evans and Kimoniset al in 1997.⁶Two major or one major and two minor criteria should be satisfied for a positive diagnosis.

The Diagnostic Criteria of Basal Cell Carcinoma Syndrome .⁶

Criteria

Major criteria

1. More than two basal cell carcinomas (BCC) or one BCC in patients <20 years
2. Histologically-proven OKCs of the jaw
3. Three or more cutaneous palmar or plantar pits
4. Bifid, fused or markedly splayed ribs
5. First degree relative with BCNS

Minor criteria

1. Proven macrocephaly, after adjustment for height
2. Congenital malformations: cleft lip or palate, frontal bossing, 'coarse face', moderate or severe hypertelorism.
3. Other skeletal abnormalities: Sprengel deformity, marked pectus deformity, marked syndactyly of the digits

4. Radiological abnormalities: Bridging of the sella turcica, vertebral anomalies such as hemivertebrae, fusion or elongation of the vertebral bodies, modeling defects of the hands and feet, or flame shaped lucencies of the hands or feet

5. Ovarian fibroma

6. Medulloblastoma

In Case 1- Our patient had multiple OKCs in both maxilla and mandible ,bifid ribs on 3rd , 4th and 5th on right side and 4th rib on left side ,frontal bossing ,hypertelorism, depressed nasal bridge ,palmar pits and sternal protuberance and in Case-2 –our patient had multiple OKCs in both maxilla and mandible , bifid ribs on 5th rib bilaterally , macrocephaly, frontal bossing ,hypertelorism , prominent supraorbital ridges, depressed nasal bridge, mandibular prognathism and plantar pits, thus suggesting to be a case of Gorlin-Goltz syndrome.

Odontogenic keratocysts appear in 75% of the syndromic patients and are normally the first symptoms. The odontogenic keratocysts in BCNS usually involve unilocular or multilocular radiolucencies of the posterior body, angle or ramus of the mandible. The lesions are often bilateral, although can be unilateral.⁷ Syndromic OKC has a higher recurrence after treatment and more aggressive behaviour than sporadic lesions.⁸ In young patients, the cysts may be associated with unerupted teeth and occasionally may cause displacement of teeth or root resorption .⁷

The management of these lesions varies in aggressiveness from simple enucleation or curettage to osteotomy with curettage of the adjacent bone.⁹ Chemical cauterization is a proved adjunctive technique in case of odontogenic keratocysts and is useful to prevent recurrence by fixing the daughter cysts or remnants of epithelial lining that are not removed during

the enucleation procedure. Carnoy's solution is a phenolic compound with tissue fixative properties. Voorsmit has demonstrated that Carnoy's solution penetrates the bone to a depth of 1.54mm following a 5 min application without any damage to the inferior alveolar nerve.¹⁰

CONCLUSION:

We must evaluate the patient with multiple odontogenic keratocysts and dental abnormalities for the risk of Gorlin-Goltz syndrome. Early diagnosis of the syndrome is essential to treat it effectively and thereby minimising complications. There should be a periodic follow-up at regular intervals of 6 months till 5 years followed by once annually for entire life.

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