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GORLIN GOLTZ SYNDROME: CASE REPORT AND LITERATURE REVIEW

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ABSTRACT
Gorlin Goltz syndrome also known as nevoid basal cell carcinoma (NBCC) syndrome is the disorder which can be inherited in a autosomal dominant way or occur due to mutation in tumour suppressor (PTCH) gene, located in the 9q22.3 chromosome. It shows varied presentations involving multiple systems of the body and showing many oral manifestations. The pathognomic clinical and radiological findings like multiple keratocysticodontogenic tumours, basal cell carcinomas, bilammelar calcification of falx cerebri, and bifid ribs can help in early diagnosis of the condition and early management preventing further complications. Here in we report a case of Gorlin Goltz syndrome demonstrating all the salient clinical and radiographic features.

KEYWORDS: Keratocystic odontogenic tumour, bifid ribs, falx cerebri calcification.

INTRODUCTION AND REVIEW OF LITERATURE

Gorlin Goltz syndrome has various synonyms like nevoid basal cell carcinoma syndrome, "basal cell nevus syndrome," "NBCCS," or the most complex name of "multiple basal epithelioma, jaw cysts bifid rib syndrome[1] and fifth phacomatosis.[2] The features of this syndrome were individually reported by various clinicians, starting with skin lesions by Jarisch and White in 1894, first documentation of the syndrome was done by Binkley and Johnson in 1951, Gorlin and Goltz accumulated the data and described the syndrome in 1960.The first historical evidence of the syndrome was found in Egypt in excavated skeletons of 11 dynasty by Satinoff and Well in 1967.[3]

Incidence of the Gorlin Goltz syndrome is estimated to be 1 in 50,000 to 150,000 in the general population. Farndon *et al.* reported a minimum prevalence of 1 in 57,000 people. Shanley *et al.* in Australia and Lo Muzio*et al*. in Italy estimated the prevalence as 1 per 64,000 and 256,000, respectively. Evans *et al.* reported that the prevalence rate in the United Kingdom was 1 per 560,000. On reviewing the literature from Science Direct and Pubmed we found that only 185 cases are reported of Gorlin Goltz syndrome from India till date. Gorlin and Goltz, in 1960, established the classical triad of this syndrome, characterized by multiple baso cellular pitheliomas, keratocysts in the jaws, and bifid ribs' which was later modified by Rayner *et al*., who established that for making the diagnosis at least odontogenic keratocysts had to appear in combination with calcification of the falxcerebri or palmar and plantar pits.[4] Along with above described major features there are many other skeletal, dermatological, neurological anamolies seen as minor manifestations in some cases.[5] Both genders are equally affected and the clinical features arise in the first, second or third decade of life.

CASE REPORT

A 16 old year female patient reported to the department of oral medicine and radiology with the complaint of pain and swelling over the right and left side of face since 3 to 4 months. Pain was continuous dull in type, low grade in intensity with accompanied swelling bilaterally more on left side of lower half of face extra-orally increasing to the present size. No history of associated fever, dysphagia and pus or watery discharge extra-orally or intra-orally. No relevant medical and dental history presented by the patient.

On examination extra-orally, there was a diffuse firm swelling, slightly tender on palpation seen on the lower half of the face bilaterally, with no overlying draining

sinus and normal temperature of the overlying skin. Lymph nodes were not palpable.

On general physical examination, the patient was moderately built nourished and presented with normal gait with satisfactory vital signs. She had a barrel-shaped chest, with drooping shoulders and mild kyphosis. On extra-oral examination, a mesoprosopic face form, concave facial profile, coarse face ,mild frontal bossing, prominent supraorbital ridges, midfacial hypoplasia, widened nasal bridge, hypertelorism, and mandibular prognathism were noted "figure 1a". Three punctuate pits were noted on the palmar surfaces of the hands[figure1c).

Intraoral examination (figure 1b) showed missing 13 14 23 27 35 37 47, buccal cortical plate expansion with respect to left side of mandible , carious 16 26 36 46, no intraoral draining sinus, over retained 75. On intraoral palpation there was expansion of buccal cortical plates with cracking sound on slight pressure application suggestive of thinning of bone.

Panoramic radiographs shows:"figure2a"

1. Well defined unilocular radiolucency with corticated borders extending from the middle third of the roots of 14, involving the impacted 13 extending upto the midline.
2. Pericoronal radiolucency seen surrounding impacted 23.
3. Impacted 28 seen.
4. Well defined multilocular radiolucency with sclerotic borders seen extending from the periapical region of 34 to the upper part of the left ramus of the mandible anteroposteriorly and extending from the alveolar border to the lower border of the mandible superinferiorly causing thinning and expansion of the lower border of the mandible and right ramus, interior of the lesion is completely radiolucent and includes impacted 35 37.
5. Well defined multilocular radiolucency with sclerotic borders seen extending from the periapical region of the 46 to the right ramus of mandible upto the right coronoid process anteroposteriorly and causing thinning and expansion of the anterior border of the right ramus of the mandible , interior of the lesion includes impacted 47.

CT scan axial view shows opacification of the left maxillary sinus (figure2b) in the antero-superior region, expansion of the left maxillary buccal cortical plate with perforation, erosion of the lateral l wall of the left maxillary sinus, expansion of the bilateral ramus of the mandible " figure 2c" perforation of the lingual border of the bilateral ramus of the mandible seen. Axial section of brain "figure 2d" shows radiopaque line running anteroposteriorly in the mid-sagittal plane suggestive of calcification of the falxcerebri.

Chest radiograph "figure 2e" shows bifid right 7th rib and bifid left 3rd rib.
Based on clinical and radiographic findings a provisional diagnosis of developmental cyst in the patient of Gorlin Goltz syndrome was made.

Surgical enucleation of the cyst was done in all four quadrants and tissue was sent for histopathological examination, which showed a cystic cavity lined by odontogenic epithelium of four- to six-cell thickness with corrugated borders and peripheral palisading nuclei. Histopathological diagnosis of keratocysticodontogenic tumour was made.

Final diagnosis of multiple keratocystic odontogenic tumor was made in the patient of Gorlin Goltz syndrome and patient was referred to the various other dental speciality for further management.
Patient was kept under 2 year follow up "figure3a and 3b"and no recurrences of the intraoral lesions were seen.

Table 1

Sr. No	Major criteria	Presented case shows
1.	Multiple (>2) Basal cell carcinoma (BCC)s or one under 20 years	No
2.	Multiple Keratocystic odontogenic tumor of the jaws proven by histopathology.	Yes
3.	Palmar or plantar pits (3 or more)	Yes
4.	Bilamellar calcification of the falxcerebri	Yes
5.	Bifid, fused or markedly splayed ribs	Yes
6.	First degree relatives with NBCCS	No

Table 2

Sr. No	Minor criteria	Presented case shows
1.	Macrocephaly determined after adjustment for height	Yes
2.	Congenital malformation: cleft lip or palate, frontal bossing, "coarse face", moderate of severe hypertelorism.	Yes
3.	Other skeletal abnormalities: sprengel deformity, marked pectus deformity, marked syndactyly of the digits.	No
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 radiolucencies of the hands or feet	No
5.	Ovarian fibroma	No
6.	Medulloblastoma.	No

PHOTOGRAPHS

Fig1: (a) Clinical profile picture of the patient showing bilateral extraoral swelling, frontal bossing and hypertelorism. (b) Intraoral picture showing expansion of the buccal cortical plates (c)Palm photograph showing palmar plantar pits

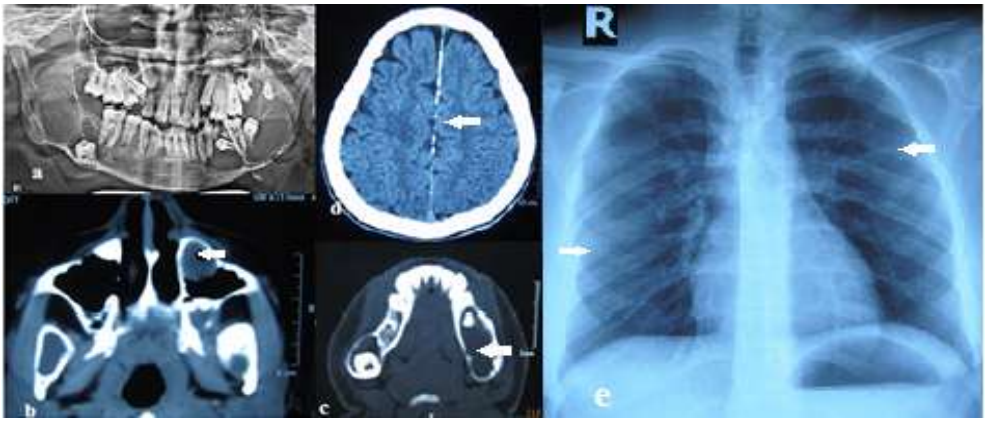

Fig2: (a) Preoperative orthopantograph of the patient showing cystic radiolucency associated with impacted teeth in all four Quadrants. (b) CT scan axial view showing opacification of the left maxillary sinus and expansion of the left maxillary buccal cortical plate. (c) CT scan axial view showing expansion of mandibular of the buccal and lingual cortical plates (d) Axial view of skull showing falx cerebri calcification. (e) Chest radiograph showing bifid ribs.

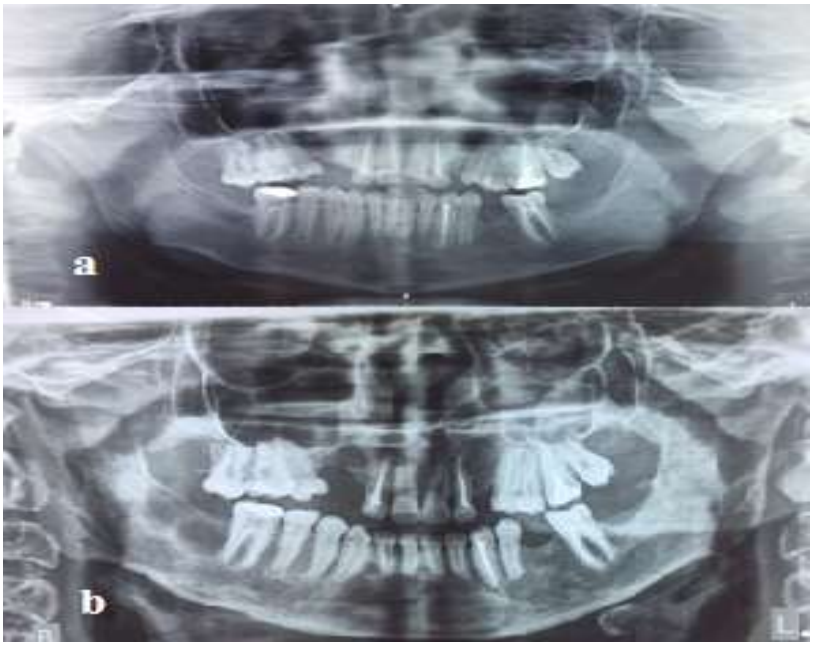

Figure3: (a) Follow up orthopantograph of 1 year
(b) Follow up orthopantograph of 2 year

DISCUSSION

Gorlin-Goltz syndrome, also known as nevoid basal cell carcinoma syndrome, comes into being due to a genetic alteration produced by a mutation in the "Patched" tumour suppressor gene, and it is inherited in a dominant autosomal way, though sporadic cases have been found. However, mutations in others genes such as Patched 2 (PTCH2), Smmothened (SMO) and Sonic hedgehog (SHH) have been reported in isolated cases of basal cell carcinoma and medulloblastoma.

This syndrome shows a high penetrance and variable expressiveness. However patient who had reported to us had no such family history of any syndrome. According to Lorenzo lo Muzio and Kimonisveetal.[5, 6] diagnosis of NBCCS can be made in the presence of two major criteria or one major and two minor criteria as shown in table 1 and 2.

Referring to the table 1 and 2 case presented in this article shows 4 major criteria and 2 minor criteria and hence was finally diagnosed as the case of Gorlin Goltz syndrome. The most common presentation of Gorlin Goltz syndrome are not life threatening, though medulloblastoma, a life threatening condition is found to be associated with 1% to 2% cases of Gorlin Goltz syndrome as reported in Evan series [8] .Early onset medulloblastoma may be the presenting sign of NBCCS; thus, in children in which this tumour is diagnosed, NBCCS should be suspected and a careful examination for other signs of the syndrome should be performed in order to rule it out.[7, 8]

The case of Gorlin Gotlz syndrome needs to be treated with different dental and medical specialities depending on the manifestation of the condition. The keratocystic odontogenic tumours needs to be enucleated with the removal of adjacent bone lining , the use of Carnoy's solution after cyst enucleation and cryosurgery is advised to prevent recurrences.[9]

CONCLUSIONS

Gorlin Goltz syndrome is a multisystemic disorder and shows varied presentation, due to its many oral manifestations, oral diagnosticians and radiologist can serve in early diagnosis of the condition and proper referral of the patient to the concerened specialist so as to prevent future complications.

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