CASE REPORT

Dentistry Section

Gorlin-Goltz Syndrome – Case Report

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ABSTRACT

The simultaneous occurrence of multiple cysts in both the jaws of a patient is rare and it usually occurs as the manifestation of a syndrome. Whenever multiple cysts are found, it is necessary to rule out an association with any syndrome, as the chances of recurrence are very high and a periodic follow-up is required for such patients. In our patient, cyst enucleation was done previously, but the association with the Gorlin-Goltz syndrome was missed and no follow-ups were advised.

KEY MESSAGE

Multiple odontogenic keratocysts which are seen in patients with the Gorlin- Goltz Syndrome have a tendency to recur and so, a periodic follow-up is necessary in such patients.

Key Words: Gorlin syndrome, Odontogenic cysts, Basal cell nevus syndrome

INTRODUCTION

Gorlin and Goltz first described the spectrum of features which are associated with the Gorlin-Goltz syndrome or the Nevoid Basal cell carcinoma syndrome (NBCCS), in 1960. It is an autosomal dominant disorder with a genetic locus on chromosome subbands and bands 9q22.3-q31, as determined with linkage analysis [1].

It is also called as the fifth phakomatosis due to the presence of multiple cutaneous, skel¬etal, ophthalmic and neurological abnormalities. It comprises of skeletal features such as the bifid rib, frontal and parietal bossing and man¬dibular prognathism and cutaneous abnormalities such as multiple basal cell carcinomas and palmar and plantar keratosis. NBCCS can also include concomitant hypertelorism, mental retardation, strabismus, calcification of the falxcerebri and medulloblastomas [2].

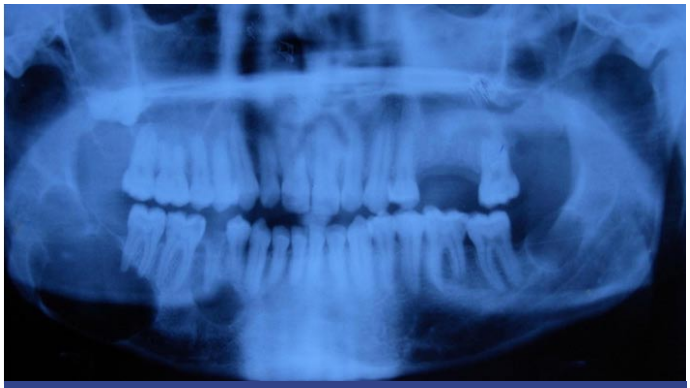
We are discussing here the possibility that the current case is an expression of NBCCS and are briefly reviewing the features of the Gorlin-Goltz syndrome.

CASE REPORT

A 35 year old male reported to the OPD of our department with the complaint of intraoral discharge in the upper right maxillary region since 3 months. The patient had a history of pain with respect to the maxillary right molars, subsequent to which extraction of the molars was done. This was followed by a discharge from the same region. The patient also gave a history of multiple jaw surgeries for cyst enucleation in the past. An intra-oral examination revealed missing 15, 16 and on pressure application, a white creamy exudate came out from that site. On general examination, the patient was found to be well built and his face showed frontal bossing, hypertelorism and prominent supra-orbital ridges [Table/Fig 1].

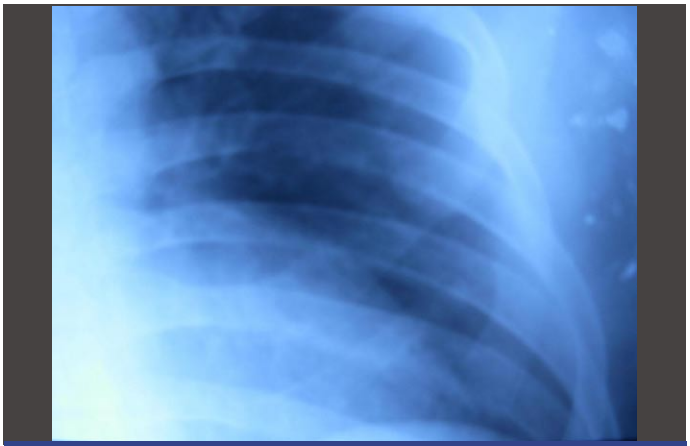
[Table/Fig 1]: Frontal Profile of patient

An Orthopantomogram (OPG) of the patient showed multiple cysts in the jaws – three in the mandible and two in the maxilla [Table/Fig 2].

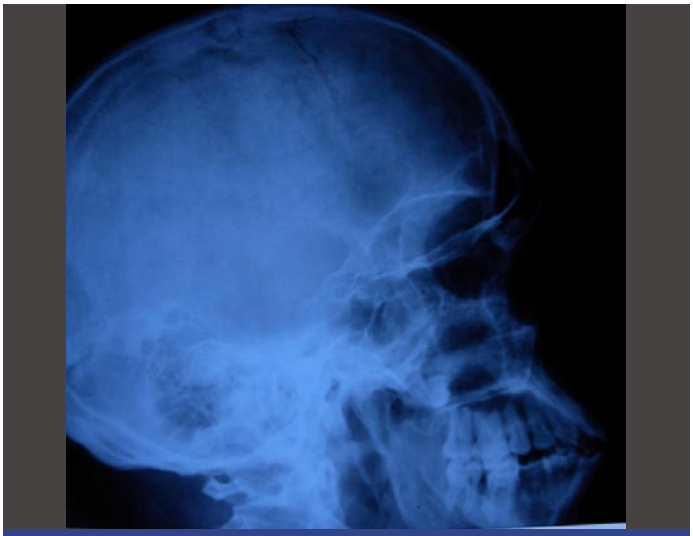


[Table/Fig 2]: Orthopantomogram showing three cystic lesions in the mandible and two involving the maxillary sinus on either side.

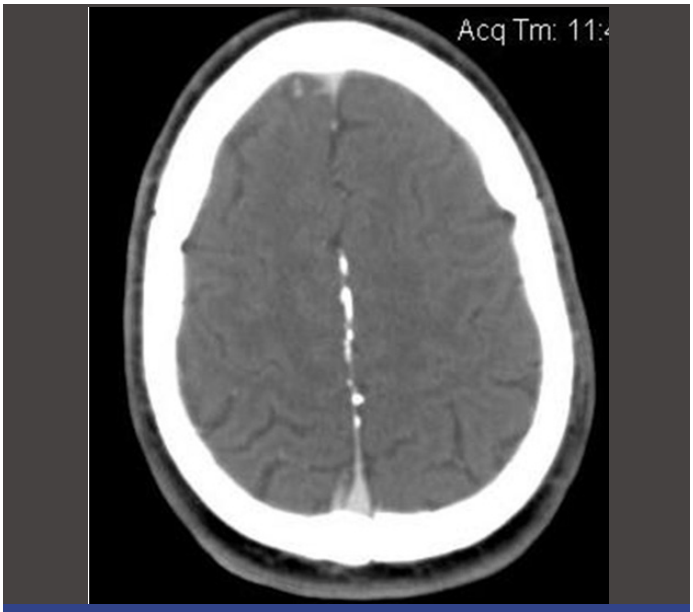
Considering the possibility of the Gorlin- Goltz syndrome, further evaluation was done with chest radiographs, which revealed a bifid rib [Table/Fig 3], lateral skull radiographs which revealed the bridging of the sellaturcica [Table/Fig 4] and CT scan of the brain which showed the lamellar calcification of the falx cerebri [Table/Fig 5].



[Table/Fig 3]: Formatted chest radiograph showing bifurcated left fifth rib

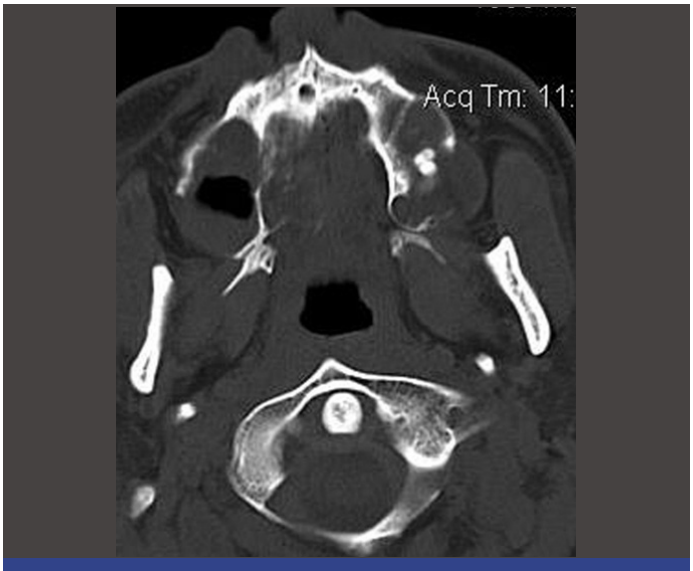


[Table/Fig 4]: Formatted lateral skull radiograph showing bridging of the sella turcica.



[Table/Fig 5]: Axial CT brain image showing falx calcification.

CT scan of the jaw of the patient revealed five expansile osteolytic lesions, two in the maxilla and three in the mandible, with thinning and breech of the cortical plates. Maxillary lesions, one on either side, were seen involving the alveolus [Table/Fig 6]



[Table/Fig 6]: Axial CT image showing expansile lytic lesions in maxilla bilaterally with cortical breech.

and were also seen to be extending into the maxillary sinus and the retrobulbar region of the orbits. An impacted tooth was seen in left maxillary lesion. On the right side, there was loss of the cortex in the maxillary tuberosity region. The mandibular lesion on the right side involved the ramus. On the left side, one lesion was seen in the periapical region with respect to the second premolar and another one was seen in the posterior part of the mandibular body, extending into the angle and the ramus. The lesions also involved the mandibular canal on the left side. Surgical enucleation of all the five cysts was done, followed by curettage. The tissues were sent for histo-pathological examination and all the cysts were found to be odontogenic keratocysts.

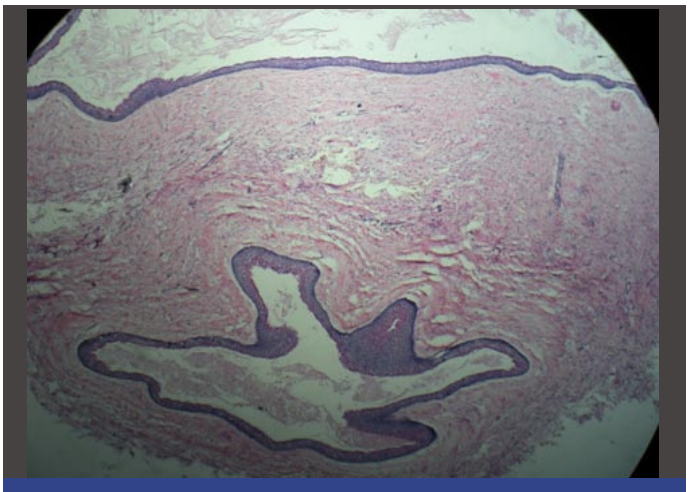
DISCUSSION

The diagnosis of the Gorlin-Goltz syndrome is made clinically by using the criteria which are suggested by Evans and others [3]. Two major or one major and two minor criteria should be satisfied for a positive diagnosis.

Our patient had three major features of NBCCS, namely bifid rib, multiple odontogenic keratocysts in the jaw and lamellar calcification of the falx and minor features such as the bridging of the sella turcica, frontal bossing, prominent supraorbital ridges and hypertelorism, thus suggesting it to be a case of the Gorlin-Goltz syndrome. Regarding the site predilection, OKCs which are associated with NBCCS are more common in the mandible with 69% involvement, as compared to 31% in the maxilla. In the mandible, 43% OKCs occurs in the molar ramus region, followed by 18% in the incisor-¬canine area. In the maxilla, 14% OKCs were found to occur in the incisor-¬canine area, followed by molar tuberosity with 12%, 7% in the mandibular premolar region and 3% in the maxillary premolar region.

Regarding the male to female ratio, it was 1:0.62 for OKCs which were not associated with NBCCS and 1:1.22 for OKCs in NBCCS. This shows that simple keratocysts are more common in males, but that more females seem to have NBCCS [4].

Based on histopathological studies, parakeratiniza¬tion, intramural epithelial remnants and satellite cysts were found to be more frequent among the OKCs which were associated with NBCCS than in the solitary keratocysts [5]. In our patient, the lining of the OKCs revealed the presence of parakeratinization and epithelial remnants in the connective tissue wall, thus indi¬cating the association with NBCCS. Multiple satellite and daughter cysts were also seen by histopathology in our patient [Table/Fig 7].



[Table/Fig 7]: Hematoxylin& Eosin (H & E) stained section shows satellite cyst.

The term “multiple cysts” does not necessarily mean that the patient must have more than one cyst at a given time; rather it refers to the occurrence of cysts over the life time of the patient [6]. Our patient also had a history of multiple cyst enucleations in the past.

There is no specific laboratory test to diagnose NBCCS, although the affected patients may have high levels of cyclic adenosine monophosphate and impaired phosphate diuresis on parathormone challenge [7].

The treatment of the Gorlin Goltz syndrome is in accordance with the generally accepted rules for the treatment of basal cell carcinomas and keratocysts in other patients. Radiation should be avoided, as it may trigger off the development of other tumors in the adjacent skin areas. Cystectomy, including the removal of the bony walls of the resulting cavity, is an adequate surgical treatment for the odontogenic keratocysts. In the treatment of the recurrent OKCs which are associated with NBCCS, the overlying surface epithelium should be excised along with the cystic lining to prevent recurrences from the residual epithelial islands and microcysts [8].

In addi¬tion, the use of Carnoy's solution following cyst enucleation (applied only over the areas where the cyst is attached to the mucosa) and cryosurgery (because of the unique ability of liquid nitrogen to devitalize the bone in situ while leaving the inorganic framework untouched) is advocated to kill the epithelial remnants and the dental lamina within the osseous structures and to thus, prevent recurrences [9].

CONCLUSION

Our case highlights the importance of the awareness of this rare syndrome, especially in young patients without any skin lesions. It is useful to keep in mind the existence of this syndrome and to recognize the presence of some major criteria that are easily recognizable in the CT scan of the head and neck, to thus establish the diagnosis, to offer the opportunity for frequent follow-ups and to therefore, increase the chances for better overall survival rates [10].

In this case, the patient had also undergone surgeries in the past for cyst enucleation, but the possibility of the Gorlin-Goltz syndrome was not considered at that time and therefore, no frequent follow-ups were advised to the patient. It is important to follow up the patients with diagnosed syndromes for the rest of their lives, because they can produce new odontogenic cysts and new basal cell carcinomas almost continuously. Basal Cell Carcinomas require frequent follow-up care, 3-4 times a year (or more), to achieve an early diagnosis and to plan the treatment. In young children who are at risk, medulloblastomas necessitate a neurological examination every 6 months, and intermittent MRIs should be considered in children who are younger than 7 years of age. Odontogenic keratocysts require dental follow-up visits, including a periodic radiographical evaluation every 6 months, especially in childhood and early adolescence [11].

Finally, the whole family of the patients with the Gorlin-Goltz syndrome should be examined and genetic counseling should be offered, as it is inherited as an autosomal dominant disorder.

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