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CASE REPORT

Clinical and Radiographic Features of Nevoid Basal Cell Carcinoma Syndrome: A Case Report

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ABSTRACT

Nevoid Basal Cell Carcinoma Syndrome, also called as Gorlin Gotz syndrome is an autosomal dominant disorder characterized by the presence of multiple odontogenic keratocysts (OKCs) of the jaw, basal cell carcinomas and skeletal anomalies. Presence of multiple OKCs is the first clinical manifestation in majority of the cases. Early diagnosis and treatment are of utmost importance in reducing the severity of this syndrome. We hereby report a rare case of Gorlin Goltz syndrome in a 28 years old female patient who presented with multiple cystic lesions in the jaws. Present case also highlights the clinical, radiographic and histopathologic features of this uncommon syndrome.

Key words: autosomal dominant, Gorlin – Goltz syndrome, odontogenic keratocyst

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INTRODUCTION

Nevoid Basal Cell Carcinoma Syndrome (NBCCS) or Jaw Cyst- Basal Cell Nevus Bifid Rib Syndrome, also known as Gorlin-Goltz syndrome (GGS) is inherited by autosomal dominant pattern and exhibits high penetrance and variable expressivity.[1,2] Jarisch and White first described it in 1894.[3] The prevalence is estimated to be 1 in 19,000 to 1 in 256,000. Frequency of occurrence is equal among males and females with sporadic and familial incidence.[1] Multiple Odontogenic keratocysts (OKCs), basal cell carcinomas (BCCs) and skeletal anomalies are the main characteristics of NBCCS.[2,3] We hereby report a rare case of GGS in a 28 years old female patient who presented with multiple cystic lesions in the jaws. Present case also highlights the clinical, radiographic and histopathologic features of this uncommon syndrome. Until date, very few cases of NBCCS are reported in India. This case report draws attention regarding multidisciplinary approach h in the diagnosis of the syndrome.

CASE REPORT

A 28-year-old female patient reported to the outpatient department with a chief complaint of pain in the lower left back tooth region for the past 4 months. The patient reported history of dull aching pain in the left lower back tooth region with history of pus discharge. Patient had consulted a local dentist where removal of teeth in the area and drainage of pus was done, and medications were prescribed. No pre-operative radiographs were taken at that time since the patient was in her 2nd trimester of pregnancy. After medical termination of pregnancy (MTP) 2 weeks later, an Orthopantomogram was taken. The patient was then informed about the presence of lesions in her jaws and was advised to seek specialty treatment at our institute. Patient also gave history of undergoing MTP one and half years ago due to foetal anomalies. Her family history was non- contributory. Extra oral examination revealed mild ocular hypertelorism with increased inner canthal distance. Intra oral examination revealed missing teeth

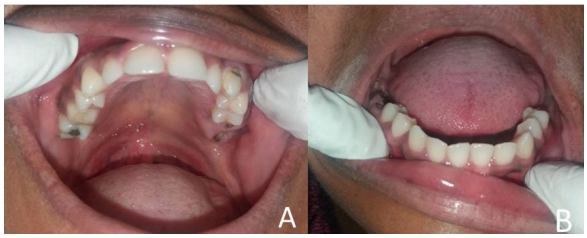

Figure 1. A&B) Intraoral photograph showing no cortical expansion in the maxilla and mandible.

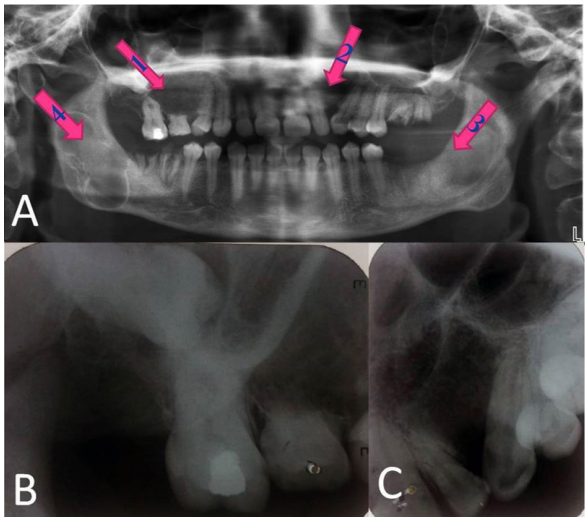

Figure 2. A) Panoramic radiograph showing multiple radiolucent lesions (arrows) in the maxilla and mandible. B) IOPA of maxillary right posterior region showing impacted tooth with presence of a pericoronal radiolucency. C) IOPA of left maxillary anterior region showing an ill-defined radiolucency causing displacement of the roots of lateral incisor and canine.

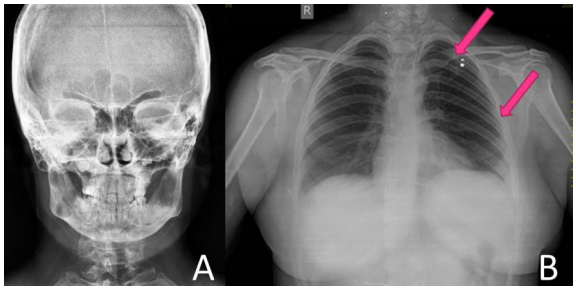

Figure 3. A) Posteroanterior cephalometric radiograph showing calcification of falx cerebri and radiolucent lesions in the mandible bilaterally. B) Chest radiograph showing mild splaying and slendering of the 2nd and 3rd ribs of the ribs on the left side.

in the region of maxillary right and left third molar, mandibular right third molar and mandibular left molars. Root stumps were noted in the maxillary left and mandibular right first and second molars (Figure 1). Panoramic radiograph showed multiple cystic lesions in all the four quadrants of the jaw. In maxilla, ill-defined radiolucent lesion was noticed on the right side in the

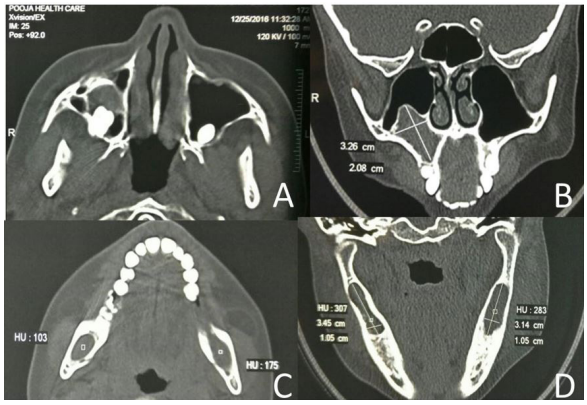

Figure 4. Axial and coronal sections showing the locations and extent of radiolucent lesions in the maxilla and mandible.

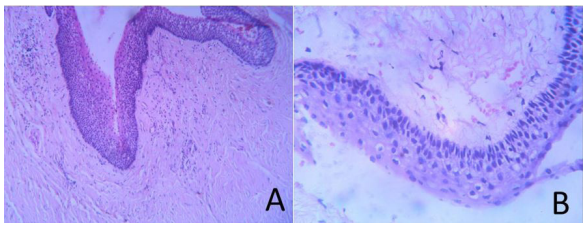

Figure 5. Photomicrograph A (10X) & B(40X): showing parakeratinised stratified squamous epithelium of 6-8 cell thickness with basal layer of palisading cells and corrugated surface.

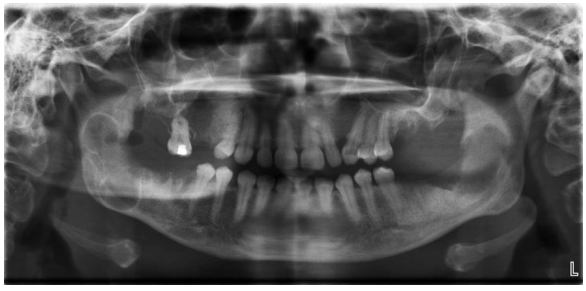

Figure 6. Post-operative panoramic radiograph.

distal aspect of maxillary second premolar extending till the crown of the impacted maxillary third molar, measuring around 31.8 x 24 mm with opacification of the right maxillary sinus and root resorption of maxillary right first molar. Another lesion was noticed in the left maxilla, an ill-defined radiolucency in the region of maxillary left lateral incisor and canine, measuring around 3.1 x 2.9 cm in dimensions with displacement of the maxillary left canine. In the mandible, a well-defined unilocular radiolucency was observed in the left molar region above the inferior alveolar canal measuring approximately 3.3 x 2.1 cms in size. Another well-defined radiolucency was noticed in the right second molar region measuring around 4.1 x 1.7 cms with scalloping margins. Loss of corticated border in the anterior aspect of the lesion was observed (Figure 2A). Intra oral periapical radiograph (IOPA) of maxillary right posterior region showed impacted tooth

with presence of a pericoronal radiolucency (Figure 2B). IOPA of left maxillary anterior region showed an ill-defined radiolucency causing displacement of the roots of lateral incisor and canine (Figure 2C). A Postero – Anterior skull view showed a radiopaque line in the midline, suggestive of calcification of the Falx cerebri (Figure 3A). Chest radiograph of the patient showed mild splaying of the ribs on the left side and the slendering of the 2nd and 3rd ribs on the left side (Figure 3B). Based on clinical and radiographic features a diagnosis of Nevoid Basal Cell Carcinoma Syndrome was made.

Surgical management of all the four cystic lesions of the jaws was done by enucleation and curettage with the usage of Carnoy's solution. The histopathological examination revealed presence of parakeratinised stratified squamous epithelium of 6-8 cell thickness with basal layer of palisading cells and corrugated surface and increased number of satellite cysts suggestive of Oodntogenic Keratocysts. Based on the clinical, radiographic, and histopathologic features diagnosis of NBCCS was confirmed.

DISCUSSION

Binkly and Johnson first described NBCCS in 1951 and later Gorlin and Goltz delineated the classical triad comprising of jaw keratocysts, multiple basal cell nevi and skeletal anomalies in 1960 that characterizes its diagnosis.[3] Later it was proposed by Rayner et al that the palmar plantar pits or calcification of the falx cerebri have to appear simultaneously along with cysts. Bettley and Ward described the association of palmar plantar pits with the syndrome.[4] Mutations of a tumour suppressor gene patched (PTCH), which is mapped to chromosome 9q22.3-q31 is considered as the underlying pathophysiology.[5] The clinical symptoms of the syndrome become apparent commonly in the 1st to 3rd decades of life.[6] In the present case the patient developed the features in the 2nd decade of life. This syndrome presents with a wide array of clinical features, the most common being the presence of multiple OKCs and numerous BCCs. Basal cell nevi when seen associated with the syndrome can arise in patients at a very young age, with development of at least one BCC by the age of 20 in 80% of these patients.[3,10] Unlike non-syndromic BCCs, these lesions can occur in both sun unexposed and exposed areas of the body, and can be a few to several hundred.[3,7] Multiple OKCs are the most common anomaly in GGS, usually appearing by the 2nd or 3rd decade of life, seen in 65–100% of patients and is mostly diagnosed as an incidental finding in routine radiographic examinations.[2,6] Present case also showed similar features. The keratocysts in the syndrome are found in both maxilla and mandible with equal predilection, when compared to non-syndromic cysts, which are mostly associated with the mandible.[2] They are frequently multiple, can be up to 10 in number. In our case 4 OKCs were found in both the jaws. The OKCs associated with the syndrome has more satellite cysts, islands of epithelial proliferation and odontogenic epithelial rests encapsulated within the cyst which is consistent with the histopathological features of the present case.[2] OKCs tend to have a high recurrence rate of 82% when seen associated with NBCCS.[2] Another common feature found in these patients is the presence of radio opaque line of falx cerebri are seen on Postero- Anterior skull view which is suggestive of the presence of bilamellar calcification. Palmar or plantar pits constitute the cutaneous manifestations of NBCCS.[2] Histologically, the pits are formed due to absence or diminution of stratum corneum layer of the epithelium.[7] Epithelial cysts, fibromas, lipomas are the other cutaneous malformations. Skeletal anomalies like splayed, synostotic, bifid, and those associated with the cervical spine are seen in about 75% of patients.[9] On the evaluation of the chest x-ray of the present case, it revealed the presence of splaying on the left side with slendering of the 2nd and 3rd ribs. Vertebral anomalies can also be present which include spina bifida occulta, scoliosis, kyphoscoliosis, and imperfect segmentation of cervical vertebrae with brachymetacarpalism.[8] Patients have characteristic facies, with temporo-parietal and frontal bossing ensuing in an increased cranial circumference (>60 cm) with eyes showing mild hypertelorism which was present in our case. Other reported oral manifestations in GGS include ameloblastomas, mandibular prognathism, cleft lip and/or palate, presence of displaced or impacted teeth and coronoid hyperplasia.[7,8]

The diagnostic criteria of GGS was established by Evans et al in 1991 and Kimoni et al in 1997,[2,3,6] where a patient should fulfil at least: two major criteria or one major and two minor criteria or one major criteria and genetic confirmation (Table 1). According to the above-mentioned diagnostic criteria, our case satisfies two of the major criteria i.e the presence of histopathologically proven OKCs and bilamellar calcification of falx cerebri and two minor criteria which were the presence of hypertelorism and splayed ribs because of which a confirmatory diagnosis of NBCCS was given.

Treatment modality of GGS varies with the clinical features present in each patient. Owing to the possibility of recurrence, patient should be provided with optimized treatment according to the clinical conditions.[4] The methods of treatment of BCC include curettage and electrodessication, cryosurgery, laser ablation, photodynamic therapy, surgical excision and ionizing radiation.[4,2,10] The treatment of OKCs is the most challenging part. The methods can be conservative, radical or aggressive. Conservative treatment such as enucleation is associated with high recurrence rate of the tumour. Better results can be obtained by the use of chemical curettage along

Table 1. Diagnostic criteria for Gorlin Goltz syndrome.[4]

MAJOR CRITERIA	MINOR CRITERIA
• Five or more BCC or one before the age of 30 • Multiple KCOTs confirmed histopathologically • Lamellar calcification of falx cerebri • Two or more palmar plantar pits • First degree relative affected	• Macrocephaly • Congenital malformation: Cleft lip or palate, frontal bossing, coarse facial features and hypertelorism • Pre axial or post axial polydactyly • Rib or vertebral anomalies – bifid, splayed, or extra ribs, spina bifida • Ovarian or cardiac fibromas • Medulloblastoma • Ocular anomalies • Pleural cysts

with enucleation. Studies have shown that the use of Carnoy's solution can reduce the recurrence rate to as low as 2%.[4] In the present case enucleation was done along with the application of Carnoy's solution in all the four lesions. Radical treatment involves the partial resection of the tumour invaded bone with a 5mm margin of healthy bone. Recurrence rate is less in this treatment modality.[3,4] The patients should be kept on a long term follow up as the OKCs associated with GGS have a high recurrence rate of 82%.[2]

CONCLUSION

Gorlin – Goltz syndrome is an uncommon condition. Accurate diagnosis of this syndrome at the earliest along with genetic assessment owing to the hereditary tendency has to be done to eliminate the possibility of family members being affected. Genetic counselling has to be given if the mother is affected with the syndrome. The patients have to be kept on a life long follow up owing to the high recurrence rate of the syndrome associated OKCs and non uniformity in the treatment modalities.

CONFLICT OF INTEREST

None declared.

REFERENCES

1. Gorlin RJ. Nevoid basal cell carcinoma (Gorlin) syndrome. Genet Med. 2004; 6(6):530-9.
2. Bree AF, Shah MR; BCNS Colloquium Group. Consensus statement from the first international colloquium on basal cell nevus syndrome (BCNS). Am J Med Genet A. 2011; 155A(9):2091-7.
3. Kimonis VE, Singh KE, Zhong R, Pastakia B, Digiovanna JJ, Bale SJ. Clinical and radiological features in young individuals with nevoid basal cell carcinoma syndrome. Genet Med. 2013;1 5(1):79-83.
4. Kiwilsza M, Sporniak-Tutak K. Gorlin-Goltz syndrome--A medical condition requiring a multidisciplinary approach. Med Sci Monit. 2012; 18(9):RA145-53.
5. Pan S, Dong Q, Sun LS, Li TJ. Mechanisms of inactivation of PTCH1 gene in nevoid basal cell carcinoma syndrome: Modification of the two-hit hypothesis. Clin Cancer Res. 2010; 16(2):442-50.
6. Hegde S, Shetty SR. Radiological features of familial Gorlin-Goltz syndrome. Imaging Sci Dent. 2012; 42(1):55-60.
7. Fujii K, Miyashita T. Gorlin syndrome (nevoid basal cell carcinoma syndrome): Update and literature review. Pediatr Int. 2014; 56(5):667-74.
8. Manfredi M, Vescovi P, Bonanini M, Porter S. Nevoid basal cell carcinoma syndrome: A review of the literature. Int J Oral Maxillofac Surg. 2004; 33(2):117-24.
9. Ramesh M, Krishnan R, Chalakkal P, Paul G. Gorlin-Goltz Syndrome: Case report and literature review. J Oral Maxillofac Pathol. 2015; 19(2):267.
10. Veenstra-Knol HE, Scheewe JH, van der Vlist GJ, van Doorn ME, Ausems MG. Early recognition of basal cell naevus syndrome. Eur J Pediatr. 2005; 164(3):126-30.

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