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Case Report

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Gorlin-Goltz Syndrome in a Healthy Elderly Patient: A Case Report

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Abstract

Gorlin-Goltz syndrome (GGS), a rare inherited autosomal dominant disorder is characterized by presence of multiple basal cell carcinomas, odontogenic keratocyst (OKCs), bifid ribs or numerous skeletal and dental anomalies. A high level of expressiveness and penetrance is shown by this entity. It is therefore, necessary to understand the major and minor diagnostic criteria for the prompt diagnosis of this condition. We hereby report a case of a 50-year-old male who presented with multiple OKCs. On carrying out further investigations, bifid ribs and calcification of falx cerebri were additional findings which lead us to the diagnosis of GGS. It is therefore necessary to consider GGS as a possible diagnosis while dealing with any case of multiple OKCs.

Keywords: Gorlin-Goltz syndrome, keratocyst, keratocystic odontogenic tumours, nevoid basal cell carcinoma syndrome, odontogenic

Introduction

Gorlin-Goltz syndrome (GGS), an inherited autosomal dominant disorder, is also known as nevoid basal cell carcinoma syndrome (NBCCS).[1] Gorlin & Goltz described the classical triad of this syndrome which is composed of multiple basal cell carcinomas (BCC), odontogenic keratocyst (OKCs) in the jaws and bifid ribs that characterizes the diagnosis of this disorder.[2] It is most found from first to third decade of life. Although all the findings are rarely seen in a single patient, this disorder affects multiple organ systems including skeletal, eye, skin, reproductive and neural.[3] The mutations in the patched tumor suppressor gene (PTCH), a human homologue of the drosophila gene mapped to chromosome 9g21-21, causes this disorder. Most mutations involve inactivating constitutional sequence variants in PTCH1, located on chromosome 9q22, while mutations in its paralog PTCH2, located on chromosome 1p34, rarely occur. The underlying basis of this disease is an abnormality in the Hedgehog (Hh) signaling pathway as suggested by genetic studies and chromosomal mappings.[4,5] The case of GGS is rarely reported in India with limited data available on its exact incidence or prevalence.[1] A 2020 case series reported only 64 cases documented in India since 1977, with just four cases showing hereditary involvement.[6] We hereby present a case of this syndrome in a 50-year-old male patient.

Case Report

A 50-year-old male patient reported to the department of Oral Medicine and Radiology with the complaints of pain and watery fluid discharge from the lower right back region of jaw for 15 days. The pain was gradual in onset, mild in intensity, dull aching and continuous. It aggravated on mastication and relieved on taking medications. The pain radiated to the right side of his upper jaw. He gave no history of swelling associated with the pain. The medical, family, and psychological history were non-contributory. He gave a history of an invasive operatory procedure with the skull (craniotomy) on right side following a head injury at the age of 10 years. There were no symptoms associated with the same.

On extraoral examination, a solitary round shaped nevus was seen on the left nasolabial fold measuring approximately 0.5 cm in size (Figure 1). An extensive bone defect was seen over the right half of forehead correlating with the history of surgery during childhood.

Intraorally, vestibular obliteration was seen in relation to the mandibular right first and second molars. Watery fluid discharge was noted from the lower posterior right back region. No tooth mobility was noted however, the alveolar mucosa adjacent to the mandibular right first premolar, second premolar, first molar, and second molar as well as the maxillary right second premolar, first molar, and second molar showed signs of inflammation and was tender on palpation. Mild expansion of the cortical plates was noted in the 1st and 4th quadrants.

An orthopantomogram (OPG) showed multiple well-defined radiolucencies in the upper and lower jaw, with the larger one extending from the periapical area of mandibular right lateral incisor to second molar with scalloped margins and evidence of thinning of inferior cortex of the jaw (Figure 2). Displacement of the inferior alveolar canal was noted inferiorly along the length of the lesion. Evidence of cortical perforation was seen in the mandibular right first premolar region. The lesion showed minimal expansion on occlusal radiograph. An ill-defined osteolytic lesion was noted with maxillary right third molar region with indistinct cortical margins and loss of density with the cortical margins of the floor of maxillary sinus. This feature was indicative of secondary infection with the lesion in the right maxillary third molar region. Another well-defined radiolucency, oval shaped, with corticated margins was noted in the periapical area of the left maxillary second molar. A small well-defined round radiolucency with corticated margins was also seen on distal aspect of the distal root of the mandibular left second molar, measuring approximately 1 cm in diameter. A provisional diagnosis of multiple OKCs was given. The differential diagnoses included GGS. A computed tomography (CT) scan of the patient confirmed the findings evident on OPG and showed calcification of the falx cerebri and the tentorium cerebellum (Figure 3) additionally. A chest radiograph of the patient showed evidence of bifid rib with the right 5th rib near the sternal end (Figure 4). Blood investigation revealed a higher parathyroid hormone level (86.2 pg/ml). Since the clinical and radiographical features satisfied 3 major criteria, a clinico-radiographic diagnosis of GGS was given with Brown's tumor as differential diagnosis.

Under general anesthesia, surgical enucleation of the

cysts was carried out along with extraction of the teeth in relation to the lesion. The specimens of the lesion from right maxilla and mandible were sent for histopathological examination.

Hematoxylin and Eosin-stained section of the specimen showed corrugated para-keratinised odontogenic epithelium with tall columnar basal cell layer. Nuclei are aligned and hyperchromatic, resembling a “picket fence” pattern. The underlying fibrous stroma is loose and collagenous. (Figure 5). All the features were suggestive of OKC. A final diagnosis of GGS was derived from clinical, radiographic and histopathological findings.

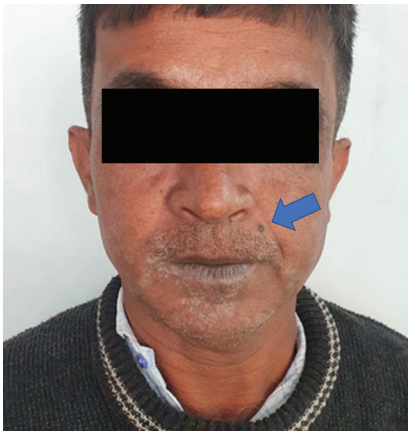

Figure 1: A solitary round shaped nevus seen on the left nasolabial fold.

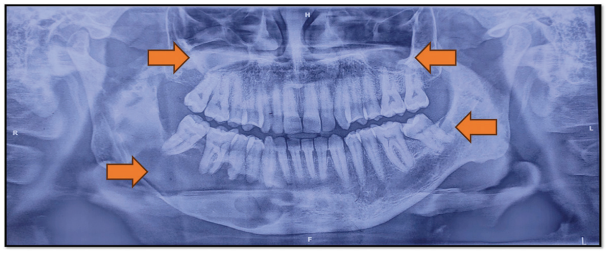

Figure 2: An Orthopantomogram (OPG) showed multiple well-defined radiolucencies in the upper and lower jaw marked with arrows.

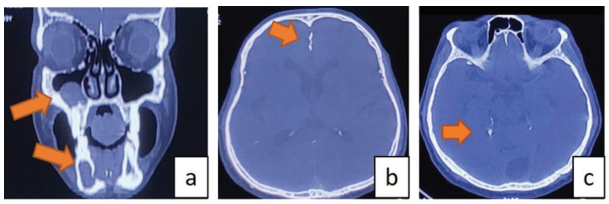

Figure 3: Computed tomography (CT) scan showing (a), Coronal view with Multiple well defined radiolucent lesions: (b), Axial view with calcification of the falx cerebri: (c), Axial view with calcification of the tentorium cerebellum.

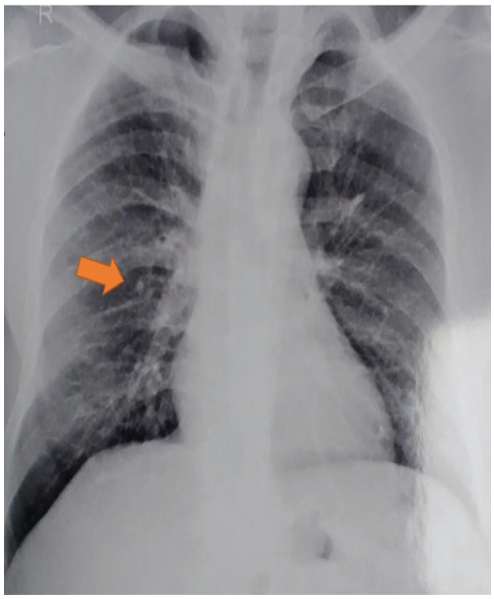

Figure 4: Chest radiograph of the patient showing bifid rib with the right 5th rib near the sterna end (Arrow).

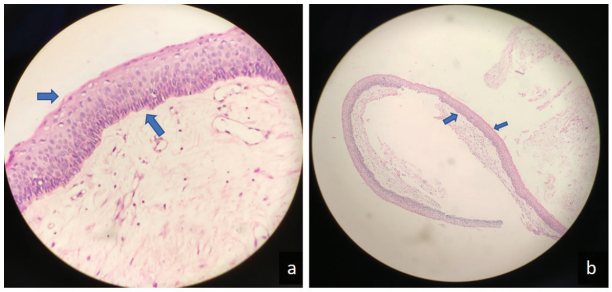

Figure 5: (a), High-power view (H&E, 100× magnification) showing the parakeratinized stratified squamous epithelial lining (indicated by the upper arrow) with a corrugated surface. The lower arrow points to the palisaded basal cell layer, composed of columnar or cuboidal cells with hyperchromatic nuclei arranged in a "picket fence" or "tombstone" pattern: (b), Low-power view (H&E, 40× magnification) illustrating the thin cystic wall and the uniform epithelial lining (arrows), which remains detached in some areas from the underlying connective tissue.

Discussion

The GGS was first reported by Jarish in 1894. Howell and Caro associated the basal cell nevus with other anomalies and disorders in 1959. Gorlin and Goltz were the first to define this condition as a syndrome.[7] Varying by the region, the incidence of this entity is 1 in 50,000-150,000 in the general population.[1] Like the present case, patients with no family history may compose 60% of total GGS patients and 35 to 50% represent new mutations.[8]

One of the most common features of 75% of GGS patients is OKC, which is often the first sign in such cases.

There is no sex predilection of OKC associated with this syndrome.[9] In a case reported by Pol *et al*.,[1] a 13-year-old boy was diagnosed with GGS based on various clinical findings including calcification of falx cerebri, frontal bossing, macrocephaly, depression of nasal bridge, hyper-telorism, prognathic mandible and palmar pits. In the present case, the patient was a 50-year-old male and showed along with multiple cystic lesions, evidence of calcification of falx cerebri and tentorium cerebellum and bifid rib. Other jaw bone abnormalities were not seen in the present case. Some other clinical findings associated with the GGS are cutaneous, dental, cardiac, sexual, craniofacial, skeletal, neurological, and ophthalmic anomalies which were not seen in the present case.[10]

The diagnosis of GGS can be made clinically by the criteria suggested by Evans *et al*., (Table 1) and modified by Kimonis *et al*. in 1997 (Table 2). For the diagnosis of GGS, two major or one major and two minor criteria should be positive.[3] Leonardi *et al*., in 2010 suggested the inclusion of atlanto-occipital ligament calcification as a diagnostic feature for NBCCS in addition to other criteria.[7] In our case three major criteria were positive namely, multiple OKCs proven by histology, calcification of falx cerebri and bifid ribs, which led us to a diagnosis of GGS.

Clinical and radiographic examination can be done to arrive at the diagnosis. However, confirmation of this syndrome is by ultrasound and DNA analysis. Diagnosis of GGS can be confirmed through DNA analysis by identifying pathogenic mutations in the PTCH1 gene, which is present in approximately 85% of affected individuals.[11] While ultrasound has limited diagnostic value, it can assist in detecting associated anomalies such as ovarian fibromas or macrocephaly, particularly in prenatal or early postnatal settings.[12]

The treatment of GGS is similar to the treatment of BCC and OKCs. Cystectomy with adjacent bony wall removal is the treatment for OKCs. Carnoy's solution can be used after cyst enucleation and cryosurgery to prevent recurrences.[13] A similar approach was followed in the present case. Treatment of BCC includes surgical excision, Mohs micrographic surgery, and for advanced or multiple lesions—as seen in GGS—hedgehog pathway inhibitors like vismodegib are effective.[14] Compared to sporadic cases, syndrome-associated BCCs and OKCs often require more conservative or repeated surgical approaches, close monitoring, and genetic counseling, as they tend to be multiple, recurrent, and aggressive.[15] Early diagnosis and treatment are important to prevent long term sequalae including

Table 1: Diagnostic criteria by Evans *et al*., in 1991.

Major criteria
More than 2 BCCs, one BCC in patients younger than 30 years of age or more than 10 basal cell nevi
Any odontogenic keratocyst (proven by histology) or polyostotic bone cyst
Three or more palmar or plantar pits
Ectopic calcification in patients younger than 20 years of age (lamellar or early falx cerebri calcification)
A positive family history of NBCC
Minor criteria
Congenital skeletal anomaly (e.g., bifid, splayed, fused or missing rib, or bifid wedged or fused vertebra)
Occipital-frontal circumference greater than the ninety seventh percentile, with frontal bossing
Cardiac or ovarian fibromas
Medulloblastoma
Lymphomesenteric cysts
Congenital malformations such as cleft lip/palate, polydactylism or eye anomaly (e.g., cataract, coloboma or microphthalmos)
BCCs: Basal cell carcinomas, NBCC: Nevoid basal cell carcinoma

Table 2: Diagnostic criteria by Kimonis *et al*., in 1997.

Major criteria
More than 2 BCCs or one BCC in patients younger than 20 years of age
Odontogenic keratocysts of the jaw (proven by histologic analysis)
Three or more palmar or plantar pits
Bilamellar calcification of the falx cerebri
Bifid, fused or markedly splayed ribs
A first degree relative with NBCCS
Minor criteria
Macrocephaly
Congenital malformations (e.g., cleft lip or palate, frontal bossing, coarse faces and moderate or severe hypertelorism)
Other skeletal abnormalities (e.g., sprengel deformity, marked pectus deformity and marked syndactyly of the digits)
Radiological abnormalities (e.g., bridging of the sella turcica, vertebral anomalies, modeling defects of the hands and feet, or flame shaped lucencies of the hands and the feet)
Ovarian fibroma or medulloblastoma
BCCs: Basal cell carcinomas, NBCCs: Nevoid basal cell carcinomas

malignancy and oro-maxillofacial destruction.

Conclusions

It can be said that GGS is an autosomal dominant genetic disorder, which is of particular interest to the dental specialists. It is a rare entity and must be considered as a possible diagnosis in all the patients with odontogenic cysts. Ignorance and delay in the management can lead to development of multiple neoplastic lesions. Early diagnosis with the help of various clinical and imaging modalities is therefore useful. Long term follow-up of such patients is also necessary to avoid development of new lesions.

Acknowledgments

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Conflicts of Interest

The authors declare that they hold no competing interests.

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