Case Report - Others

Multiple keratocystic odontogenic tumors in nevoid basal cell carcinoma syndrome

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ABSTRACT

Keratocystic odontogenic tumor (KCOT) is of particular interest because its recurrence rate is high and its behavior is aggressive. Nevoid basal cell carcinoma syndrome (NBCCS), which is also known as Gorlin syndrome, is a hereditary condition characterized by a wide range of developmental abnormalities and with a predisposition to neoplasms. These multiple KCOTs have warranted an aggressive treatment at the earliest because of the damage and possible complications. Recurrence of these lesions is a characteristic feature that has to be considered while explaining the prognosis to the patient. Here, we report a case of a 14-year-old boy with clinical features of basal cell nevus syndrome and multiple KCOTs. In addition to the other common features, congenitally missing third molars in all the four quadrants is a feature which has not been previously reported in association with NBCCS in Indian patients.

Keywords: Gorlin–Goltz, keratocystic odontogenic tumor, neoplasm, nevoid basal cell carcinoma

INTRODUCTION

The nevoid basal cell carcinoma syndrome (NBCCS) is a generalized disorder, which consists principally of multiple nevoid basal cell carcinomas (BCCs), keratocystic odontogenic tumors (KCOTs) of the jaws, vertebral and rib anomalies, and intracranial calcifications.[1] Earlier description of this condition has been mentioned in literature, but the syndrome was clearly delineated by Gorlin and Goltz in 1960.[2] KCOTs associated with NBCCS which occur earlier in life exhibit a greater tendency to recur and are more aggressive than the nonsyndromic KCOTs,[3] and they have occasionally been reported to transform into aggressive neoplasms such as ameloblastomas and squamous cell carcinoma.[4]

Occurrence of multiple KCOT is rare and there have been only 23 cases of NBCCS reported in Indian patients in the medical literature over a period of 37 years (1977–2014).[5] Here, we report a case of multiple KCOTs involving the maxillary jaw in a 14-year-old patient with bilamellar calcification of the falx cerebri along with palmar and plantar pits, thus presenting an expression of NBCCS.

CASE REPORT

A 14-year-old male patient presented with his parents with a chief complaint of pain and swelling on the right side of the face since 1½ month (an informed consent from his parents was obtained). His medical and dental history was unremarkable. On examination, he was thin built. There was an obvious hypertelorism along with fused eyebrows and an increased intercanthal distance. His gait was normal. An extraoral swelling was present on the right side of the face extending from the medial acanthus of the eye to the corner of the

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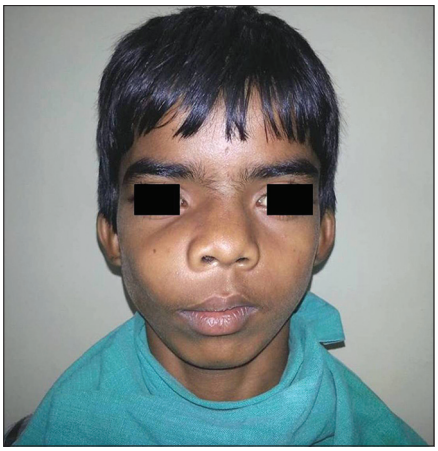

Figure 1: Extraoral photograph of the patient showing a swelling on the right side of the face

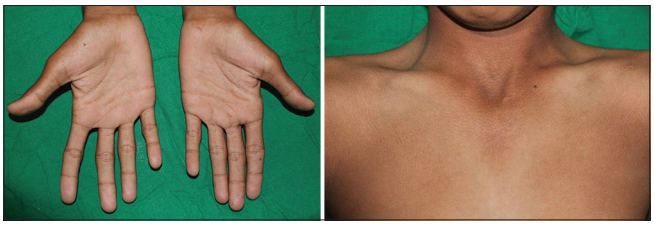

Figure 3: Palmer pits and nevi on the chest

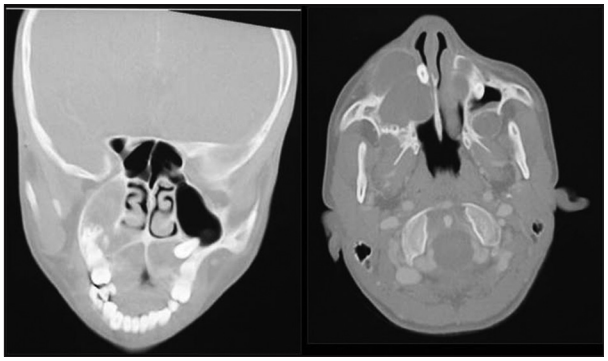

Figure 5: Computed tomography coronal and axial sections showing well-defined corticated hypodense expansile lesions involving right and left side of the maxilla

mouth, obliterating the nasolabial angle [Figure 1]. On intraoral examination, a vestibular swelling was observed which was firm, tender, and nonfluctuant extending from the deciduous maxillary right canine to the permanent maxillary first molar region. Marked palatal expansion was also noted bilaterally extending from the central incisor to the second premolar on both sides and meeting at the midline [Figure 2]. Moreover, he exhibited multiple palmar and plantar pits along with nevi on the chest and back [Figure 3].

Radiological investigations performed for the patient were orthopantomogram, occlusal radiograph, chest radiograph, and computerized tomography (CT).

Panoramic radiography revealed two large bony defects showing a large radiolucency with sclerotic borders

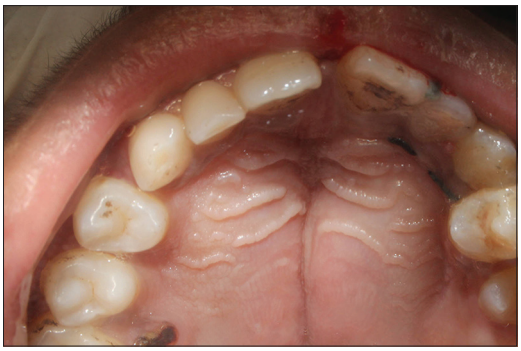

Figure 2: Intraoral photograph of the patient showing a bilateral palatal swelling

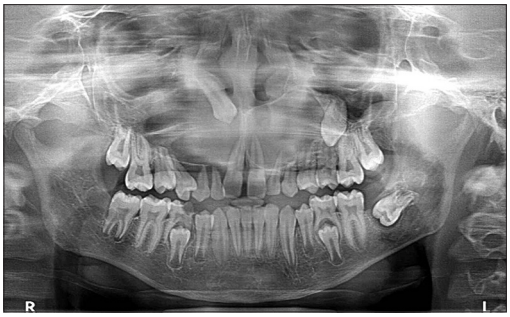

Figure 4: Orthopantomogram showing bilateral radiolucent lesions in the maxilla

resembling cysts associated with impacted permanent maxillary right and left canine [Figure 4]. The permanent maxillary and mandibular third molars were congenitally absent in all the four quadrants.

Axial and CT coronal sections showed the presence of well-defined corticated hypodense expansile lesions involving the right and left side of the maxilla. The right side showed displacement of the lateral wall of the nose and floor of the maxillary sinus superiorly. Soft tissue window of the coronal CT showed calcification of falx cerebri [Figures 5 and 6]. The chest radiograph was unremarkable.

Hematological investigations were within normal limits. Histopathological examination of the incisional biopsy confirmed the presence of KCOT. Under general anesthesia, the patient underwent marsupialization for the lesion in the right and left maxillary canine region. Enucleation and curettage with the use of Carnoy's solution and a bismuth iodoform paraffin paste pack placement were carried out along with the removal of impacted right and left canine [Figure 7]. All the lesional tissues were sent for histopathological examination. Follow-up after 1 month showed good healing at the surgical site without any paresthesia. The patient is under follow-up since the last 1 year.

Hematoxylin and eosin stained sections of both the lesions showed the classical feature of parakeratinized KCOT with a uniform 6–8 cell layer thickness. The lining epithelium showed well-defined columnar basal cells with a palisaded arrangement and with polarized nuclei. Satellite cysts and

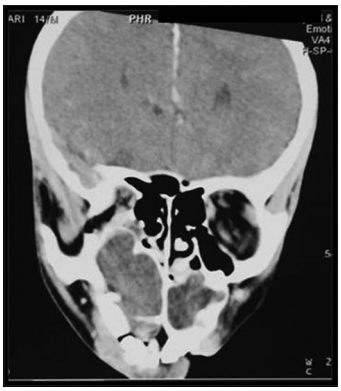

Figure 6: Soft-tissue window of coronal computed tomography showing calcification of falx cerebri

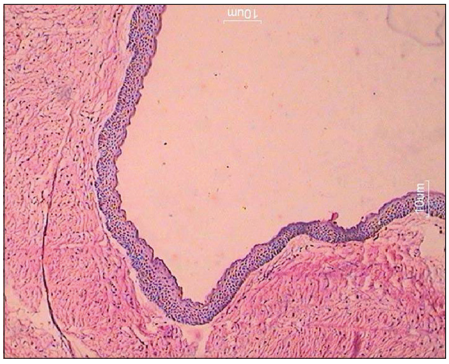

Figure 8: Hematoxylin and eosin stained section showing typical palisading arrangement of basal cells of odontogenic keratocyst lining epithelium (H and E, ×10)

epithelial remnants were not observed in the connective tissue capsule [Figure 8].

DISCUSSION

The biologic behaviors of KCOTs which are associated with NBCCS are more aggressive, and these cysts have higher recurrence rates (82%) compared with solitary keratocysts (61%).[6] There is no specific test which can diagnose NBCCS, and the diagnosis was made clinically using the criteria suggested by Evans *et al*.[7] and Kimonis *et al*. [Table 1].[8]

Some studies have shown that the affected patients with NBCCS may have high levels of cyclic adenosine monophosphate and impaired phosphate diuresis on parathormone challenge.[9] Skin involvement with multiple BCCs and palmar-plantar pits is seen in only 43% of Indian patients.[5] However, in the present case, the patient was apparently healthy and only had multiple KCOTs, palmar-plantar pits along with bilamellar calcification of the falx cerebri.

KCOTs of the jaws have been considered as developmental cysts and account for 10–15% of all jaw cysts that arise from cell rests of the dental lamina and occur over a wide age group with a peak incidence in the second and third decades of life.[10] In the present case, the patient was in the second decade. Based on histopathologic studies, parakeratinized epithelium, intramural epithelial remnants, and satellite cysts are seen more

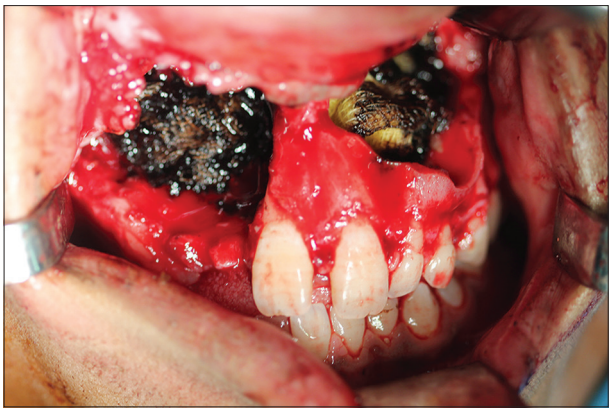

Figure 7: Enucleation and curettage of the lesion along with placement of a bismuth iodine paraffin paste pack

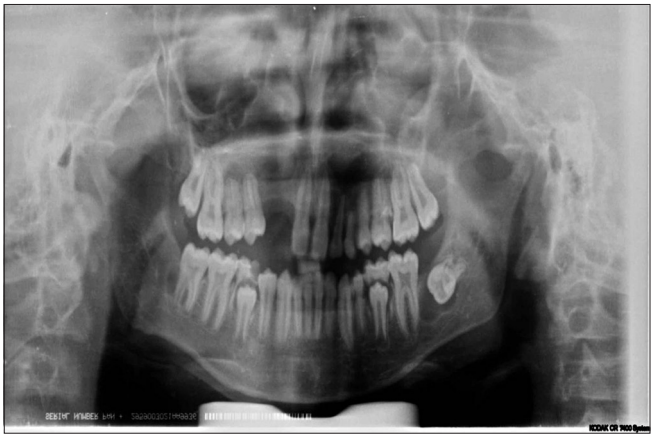

Figure 9: Postoperative orthopantomogram after 1 year showing the occurrence of a new cyst in the mandibular left third molar region

frequently among KCOTs associated with NBCCS than in solitary keratocysts.[6] In the present case, the lining of the KCOT revealed a parakeratinized epithelium in both the cysts. However, satellite cysts were not seen in the present case. Immunohistochemical studies have shown that cytokeratin (CK17) and CK19 are over expressed in KCOTs, suggesting that this may be a valuable additional parameter to distinguish between KCOT and other odontogenic cysts.[11] CK expressions were not investigated in the present case.

Once the diagnosis of NBCCS is made, then screening for the syndrome must be carried out in other family members and genetic counseling must be offered.[5] In the treatment of KCOTs associated with NBCCS, overlying surface epithelium has to be excised along with the cystic lining to prevent recurrences from residual epithelial islands and microcysts.[12] The use of Carnoy's solution following cyst enucleation and cryosurgery is advocated to kill epithelial remnants and dental lamina within the osseous margin thus preventing recurrences.[13] In the present case, the two cysts were enucleated under general anesthesia, and Carnoy's solution was applied to the osseous margins. The patient was followed regularly and after 12 months of treatment had no symptoms of recurrence of cysts. However, the patient showed the occurrence of a new cyst in the mandibular second molar region on the left side for which he is to be treated [Figure 9].

Table 1: Diagnostic criteria for nevoid basal cell carcinoma syndrome

According to Evans *et al.*	According to Kimonis *et al*.
Major criteria	Major criteria
Multiple BCC or one occurring under the age of 20 years	More than 2 BCC or 1 BCC in a patient <20 years of age
Histological proven KCOTs of the jaws	KCOTs of the jaws (proven by histopathologic analysis)
Palmer or planter pits (three or more)	Three or more palmar or plantar pits
Bilamellar calcification of the falx cerebri	Bilamellar calcification of the falx cerebri
Bifid, fused or markedly splayed ribs	Bifid, fused or markedly splayed ribs
Family history of NBCCS	A first-degree relative with NBCCS
Minor criteria	Minor criteria
Macrocephaly	Macrocephaly
Congenital malformations such as cleft lip or palate, frontal bossing, polydactylism, or eye anomalies	Congenital malformations such as cleft lip or palate, frontal bossing, coarse facies and moderate or severe hypertelorism
Other skeletal abnormalities such as bifid rib, sprengel deformity, and marked pectus deformity	Other skeletal abnormalities (e.g., sprengel deformity, marked pectus deformity and marked syndactyly of the digits)
Radiological abnormalities such as bridging of sella tursica, vertebral anomalies	Radiological abnormalities (e.g., bridging of the sella turcica, vertebral anomalies, modelling defects of the hands, and feet flame-shaped lucencies of the hands and feet)
Ovarian fibroma	Ovarian fibroma or medullablastoma (not applicable if patient is male)
Medulloblastoma	
Lymphomesentric cysts	

NBCCS=Nevoid basal cell carcinoma syndrome; BCCs=Basal cell carcinomas; KCOT's=Keratocystic odontogenic tumors

Clinical and radiological criteria will continue to play an important role in the diagnosis of NBCCS. Many newer findings are being added to the spectrum of the clinical and radiological manifestations in NBCCS, such as bilateral coronoid hyperplasia, sloping shoulders, low-pitched voice, supernumerary teeth, Talons cusp, just like we found congenitally missing third molars in all four quadrants.[5] In conclusion, for any patient with multiple KCOTs, the possibility of NBCCS must be considered. A complete clinical examination and histopathological analysis must be performed to detect any features associated with this syndrome. As KCOTs may be the first and only manifestation of NBCCS, the dentist may be the first to detect it. This case emphasizes the need for a thorough examination of the patient since KCOT associated with NBCCS have a high recurrence rate.[6] The present case thus adds to the other numbers of such cases of syndromic KCOTs. It is important that we highlight the importance of diagnosing this entity so that a stringent follow-up may be enforced in such cases.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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