CASE REPORT

Diagnosis of Jaw Cyst Basal Cell Nevus Syndrome from Multiple Odontogenic Keratocysts in a 3D CT Scan

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ABSTRACT

The purpose of this report is to discuss the diagnosis of Jaw cyst basal cell nevus syndrome. Two components of the syndrome are multiple odontogenic keratocysts and basal cell carcinomas. The discovery of multiple keratocysts is usually the first manifestation of the syndrome. In the case discussed, 3D CT scan taken to evaluate a diagnosed odontogenic keratocyst revealed more than one keratocysts. This led to the closer evaluation and diagnosis of Jaw cyst basal cell nevus syndrome.

Keywords: Basal cell nevus syndrome, Jaw cyst basal cell nevus syndrome, Odontogenic keratocyst, 3D CT scan.

INTRODUCTION

Odontogenic Keratocysts (OKCs) are developmental odontogenic cysts of epithelial origin, first identified and described in 1876 and further characterized by Phillipsen in 1956. Odontogenic keratocyst (OKC) is known for its high recurrence rate, aggressive behavior and its association with Jaw cyst basal cell nevus syndrome. Jaw cyst basal cell nevus syndrome or Gorlin goltz syndrome involves multiple organ systems. A fully expressive case presents with several of the many possible abnormalities of skeletal, skin, eye, reproductive and neural system. However, a subtle case presents with only one or two abnormalities that may not be even very obvious. Multiple OKCs is one of the two most common physical findings of Jaw cyst basal cell nevus syndrome, the other being basal cell carcinomas. Presence of multiple OKCs in the jaw alerts, the clinician to the possibility of jaw cyst basal cell nevus syndrome.

The following case report, discusses a patient, diagnosed with odontogenic keratocyst of the mandible, who, on further investigation with a 3D CT scan, revealed multiple OKCs leading to subsequent diagnosis of jaw cyst basal cell nevus syndrome. The case report also illustrates the need for increase awareness of the various signs and symptoms and the use of simple diagnostic tests to diagnose the syndrome as all cases may not have all the abnormalities.

CASE REPORT

A 24-year-old male was referred to department of oral and maxillofacial surgery, Army College of Dental Sciences, Secunderabad, from a district hospital with the chief complaint of swelling in the mandible for one and half years. The patient had noticed an increase in size of the swelling for the past 6 months.

The patient was moderately built with normal gait. Facial examination revealed mandibular asymmetry and confluence of eye brows. Extraoral swelling in the mandibular region extended from the ramus on one side to the other (Fig. 1). A depression was observed before the left ramus. The swelling was not fluctuant and not tender on palpation.

Radiographic examination, using orthopantomogram, revealed a multilocular lesion involving the right ramus, body of mandible and extending to the other side of mandible without involving the left ramus. The upper left third molar was impacted and there were no cystic changes apparent on the panoramic radiograph (Fig. 2).

Blood investigations including CBP, serum calcium, phosphorus and alkaline phosphates were within normal limits.

Aspiration biopsy was positive with yellow liquid aspirate.

Incision biopsy was suggestive of odontogenic keratocyst.
A 3D CT scan was done for evaluation of lesion before surgery revealed an extensive cyst associated with the impacted, upper left third molar, obliterating the left maxillary sinus and a small developing lesion in association with upper right third molar (Fig. 3).

The presence of these multiple cysts led to further investigations and closer evaluation of the patient. Increased cranial circumference of 59 cm (> 55 cm), pectus excavatum, nevi on the chest were seen (Figs 4 and 5). Skull X-ray revealed calcification of falx cerebri (Fig. 6). The clinical and radiological findings were confirmatory for the Jaw cyst basal cell nevus syndrome.

Under general anesthesia enucleation of mandibular and maxillary odontogenic keratocysts was performed, followed by chemical cauterization of cavity for 3 minutes with Carnoy’s solution. The upper right second and third molars, upper left second and third molars, lower right second and third molar teeth were extracted. Cystic lining was sent for histopathological examination and the report confirmed the diagnosis of odontogenic keratocyst, the upper right maxillary lesion was close primarily the upper left maxillary and the mandibular lesion was left open to oral cavity with regular irrigation and packing with chlorhexidine tulle packs.

The patient has been on regular follow-up since the surgery. One postoperative X-ray showed good healing.

**DISCUSSION**

Jaw cyst basal cell nevus syndrome is an ectomesodermal polydysplasia with numerous manifestations, characterized most often by (a) cutaneous abnormalities, including multiple basal cell abnormalities, benign dermal cysts, palmar-plantar pits; (b) craniodentofacial anomalies, such as OKCs, malocclusion, broad nasal bridge and increased head circumference; (c) skeletal anomalies, including frontal and parietal bossing and mandibular prognathism as well as costal anomalies involving the rib and vertebrae; (d) ophthalmologic abnormalities, including hypertelorism, congenital blindness, and strabismus; and (e) neurologic anomalies, including calcifications of the falx cerebri, bony bridging of the sella turcica and medulloblastoma.¹

The components of Jaw cyst basal cell nevus syndrome divided into two groups, as suggested by Evan et al⁴ and Kimonis et al⁵ with so-called major and minor criteria. The final diagnosis of the syndrome is achieved upon finding any two of the major criteria, or one major criterion plus two minor criteria.
Multiple OKCs are a well-recognized feature of Jaw cyst basal cell nevus syndrome. Usually, OKC development, which comprises one of the major criterions, occurs before the observation of other clinical findings in Jaw cyst basal cell nevus syndrome, enhancing early diagnosis of the disease. OKCs associated with this syndrome have a familial tendency and early family detection and genetic counseling are critical. These cysts arise earlier in patients with Jaw cyst basal cell nevus syndrome than in those who are unaffected by the syndrome.

OKCs associated with Jaw cyst basal cell nevus syndrome have occasionally been reported to transform into aggressive neoplasms, such as ameloblastomas and squamous cell carcinoma. The cyst lining seen in the Jaw cyst basal cell nevus syndrome-related OKC is classically parakeratinized and does not appear to be associated with the orthokeratinized variant of the OKC.

The reported frequency of recurrence of the odontogenic keratocyst ranges from 2.5 to 62.5% in various studies. Regezi pointed out that the recurrence rate of solitary OKC is 10 to 30%. Approximately, 5% of patients with odontogenic keratocysts have multiple sporadic jaw cysts (nonsyndromic) and that their recurrence rate is greater than that for solitary lesions.

OKC can be managed by enucleation and curettage surgery. McIntosh has advocated the resection of odontogenic keratocyst with 5 mm linear margins as the preferred primary method of treatment. Vedtofte and Praetorius advocated the excision of overlying mucosa in conjunction with the removal of the cyst. Williams and Connor recommended a primary enucleation and curettage surgery for odontogenic cysts including the use of methylene blue as a marking agent, followed by application of Carnoy’s solution for 3 minutes. Treatment by resection is advocated for recurrent lesions. Resection is not warranted in syndromic patients, marsupialization is more desirable.

CONCLUSION

Jaw cyst basal cell nevus syndrome can present with several of the many possible abnormalities that may not even be very obvious in a subtle case. The clinician is usually alerted to the possibility of Jaw cyst basal cell nevus syndrome when more than one odontogenic keratocyst is found in the jaw, or when an young individual develops basal cell carcinomas. Most often patients come with a complaint of jaw swelling to the dentist. Increased awareness of the various signs and symptoms will lead to early diagnosis of this familiar syndrome. This case report has shown the superiority of a CT scan in diagnosing small developing and multiple lesions not apparent on the panoramic radiograph.

REFERENCES


