

Gorlin's syndrome presenting as recurrent mandibular cyst infection

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Abstract

A 38 year-old man with a history of painful swelling in the left mandibular angle region and skin lesions on the upper chest is reported. The symptoms, findings, diagnostic criteria and the approach for treatment are reviewed. Histological examinations of mandibular cystic and skin nodular lesions were reported as "Inflamed Keratinous Cyst" and "Basal Cell Carcinoma". It is important to consider the syndrome encountering multiple jaw cysts because of the high recurrence rate, associated medical problems, and need for close life-long follow-up of this syndrome.

Keywords: Mandibular cyst; Gorlin's syndrome; Basal cell carcinoma

Introduction

Gorlin's or nevoid basal cell carcinoma syndrome (NBCCS) is an autosomal dominant disorder, whose mutated gene is located on the long arm of chromosome.¹ The incidence of this syndrome is approximately 1 per 60,000. About one-third of cases occur in patients with no family history of the syndrome. Patients most commonly present in the third or fourth decade of life with either dental cysts or basal cell carcinomas.² They rarely present with medulloblastoma in childhood. In a significant percentage of patients, the syndrome is never diagnosed.³

Case Report

A 38 year-old male patient presented with a history of painful swelling in the left mandibular angle region and halitosis. There were no chills, fever, dyspnea, dental pain or facial trauma. He had a history of severe swelling in the same region two years before and was treated by intravenous antibiotics. His family

history was negative for similar symptoms. Physical examination revealed a nearly 5×5 cm firm, tender, fixed mass with ill-defined margins in the left side of the mandibular region with bulging of the buccal surface of the left side of the mandible and extension to the ipsilateral cheek. Dental occlusion was obviously altered, and the left mandibular teeth were deviated to the right. Parotid examination was normal. In the needle aspiration of the inflamed region, about 20 ml malodor, brown liquid (pus) was aspirated. Interestingly, panoramic x-ray revealed multiple mandibular cysts with simultaneous three radiographic forms of odontogenic ones: follicular cyst associated with the completed crown of an unerrupted or impacted tooth, radicular cyst associated with the tooth root, and perimordial cyst arising where there is no associated apparent tooth formation (Figure 1). Additionally, CT scan of the face revealed a right maxillary bone cyst (Figure 2). So with the diagnosis of abscess formation, the patient was admitted emergently and intravenous antibiotics were started. He underwent surgery and through a Risodon incision of the left mandibular angle and ramus, enucleation and curettage of the cysts were performed. No difficulty was encountered intra-operatively and postoperative recovery was uneventful except for another episode of mild swelling in the incision line, which was treated by incision and drainage.

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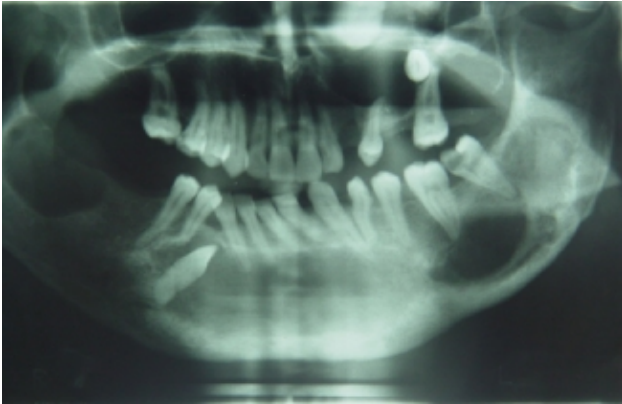


Figure 1: Panoramic X- Ray: Three types of odontogenic cysts are seen.

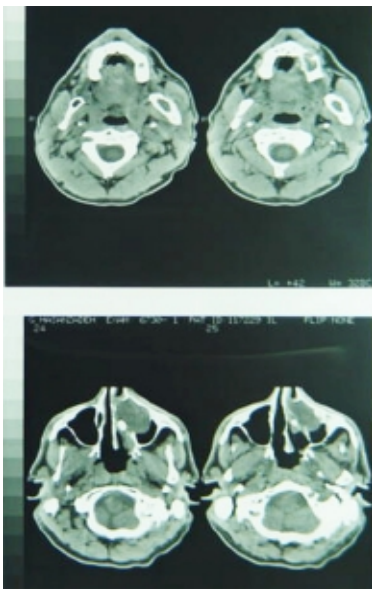


Figure 2: Facial CTscans. Right large maxillary bone cyst & multiple mandibular cysts are seen.

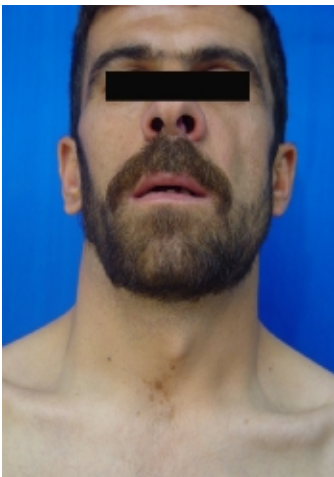


Figure 3: Skin lesions on the lower neck and upper chest.

Histological examination of surgical specimens confirmed the diagnosis of inflamed keratinous cyst. Surveillance for other components of this syndrome revealed fine pale brown lesions on the upper chest and lower neck (Figure 3). Excisional biopsy of these lesions confirmed histological diagnosis of basal cell carcinoma. Calcification of the falx cerebri was found on brain CT scan. Radiographic studies of the chest, abdomen and pelvis were normal.

Discussion

Odontogenic keratocysts in Gorlin's syndrome are found in 90% of those over 40 years of age and in 80% of those over 20. The average number of cysts is 5 but can range from 1 to 30. Those in the mandible are three times as common as those in the maxilla. Although the cysts may be extremely large, they rarely cause symptoms but markedly affect tooth displacement. Jaw fractures almost never occur. About one-half present with swelling, a quarter with mild pain, and 15% with unusual taste following the rupture of a cyst.⁴ In our patient, infection of one of the mandibular cysts was the presenting sign. Kimonis (1997) has proposed that the diagnosis of Gorlin's syndrome can be made in the presence of two major or one major and two minor criteria.⁵

The major criteria are two or more basal cell carcinomas in a person younger than 20 years of age, odontogenic keratocysts of the jaw, three or more palmar or plantar pits, bilamellar calcification of the falx cerebri, bifid, fused, or markedly splayed ribs and first degree relative with Gorlin's syndrome. The minor criteria are macrocephaly (a relative macrocephaly is found in 50%),⁴ congenital malformations (e.g. cleft lip or palate, frontal bossing, coarse face, hypertelorism), other skeletal abnormalities, such as Sprengel deformity (10%-40%),⁴ marked pectus deformity or syndactyly of finger 2 to 3, radiographic abnormalities such as hemivertebrae, fusion or elongation of the vertebral bodies, modeling defects of the hands and feet, or flame-shaped lucencies of the hands or feet, ovarian fibroma (fibrosarcoma and primary ovarian leiomyosarcoma have also been reported),^{6,7} and medulloblastoma (the next most common brain tumor is meningioma, although infrequent).³

It is cited that the incidence of several other neoplasms might also increase in this syndrome (e.g. leiomyosarcoma, thyroid adenoma, melanoma, ameloblastoma, Hodgkin's lymphoma, non-Hodgkin's

lymphoma, rhabdomyosarcoma, nasal dermoid, shwanoma, pleiomorphic adenoma of parotid, adenoid cystic carcinoma of salivary gland).^{1,8,9} Our patient had three major criteria of Gorlin's syndrome; therefore, the diagnosis was confirmed. Standard treatment for odontogenic keratocysts (presenting symptom of this patient) is enucleation and curettage.¹⁰ A recurrence rate from 10% to 60% has been reported.^{4,10} Although our patient was a recurrent

case, he has had no more problems in these three years of follow up. It is important to distinguish the odontogenic keratocyst in Gorlin's syndrome from other odontogenic cysts because of the high recurrence rate and the other medical problems that like other "cancer syndromes" might accompany the syndrome. Therefore, a close life-long follow-up of the patients is required.

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