CASE REPORT
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Maxillomandibular Cysts and Multiple Basal Cell Carcinomas Due to Gorlin-Goltz Syndrome: A Case Report

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ABSTRACT

Maxillomandibular cysts may be encountered frequently and they may be the first signs of various syndromes. In Gorlin-Goltz syndrome, skeletal, ocular and dermal lesions may accompany the maxillomandibular cysts. In order to reach a diagnosis, a thorough evaluation and a multidisciplinary approach are required. In this study, a patient with multiple maxillomandibular cysts and multiple basal cell carcinomas was presented.

Keywords: Basal cell carcinoma; Gorlin-Goltz syndrome; maxillomandibular cyst; odontogenic keratocyst

INTRODUCTION

Gorlin-Goltz syndrome is a rare autosomal dominant genetic disorder which involves dermal and skeletal anomalies (1). This syndrome is also referred as nevoid basal cell carcinoma syndrome (NBCCS) (1). It is mostly characterized by basal cell carcinomas (BCC), odontogenic keratocysts (OKC), palmar and plantar pitting, facial dysmorphism and opthalmologic and skeletal abnormalities (1). Diagnosis of the patients is based on clinical, radiological and histopathological findings.

NBCCS has a prevalence of 1 in every 56,000 to 256,000 individuals (2). A classical triad is described by Robert James Gorlin and Robert William Goltz, which consists of multiple basal cell carcinomas, jaw cysts and skeletal abnormalities (2). In addition, many other clinical manifestations are present in NBCCS. In order to reach a diagnosis, major and minor diagnostic criteria should be fulfilled (3). Due to the complexity of clinical manifestations, a multidisciplinary approach is necessary for diagnosis and follow-up (4).

CASE PRESENTATION

In January 2022, a 19 year-old female patient was referred to a dental clinic with mandibular swelling. Due to the presence of multiple maxillomandibular cysts at the panoramic graph, the patient was consulted to the Plastic Reconstructive and Aesthetic Surgery Department (Figure 1). The patient did not report any relevant personal or familial history.

In physical examination, mandibular swelling was seen by both intraoral and extraoral examination. In fact, both hemimandibles and left hemimaxilla were affected. Poor dental hygiene was seen during the intraoral examination, and bilateral lower gingivobuccal sulci and left hemimaxilla were expanded. These swollen areas were solid and expansile and

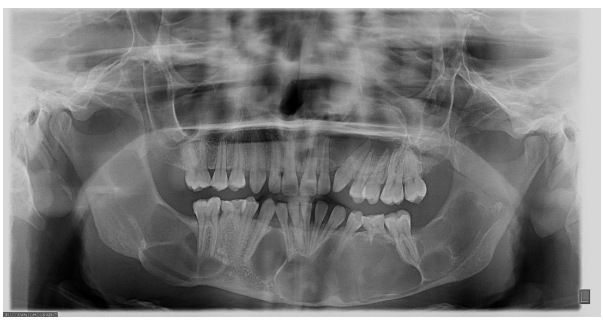

Figure 1: Preoperative panoramic radiograph of the patient is demonstrated.

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there was not any pulsation over the lesions. A computerized tomography was performed in order to confirm the localization of the cysts (Figure 2).

After further evaluation, excisional biopsy was performed under general anesthesia. During the routine follow-up of the patient, multiple pigmented lesions were detected on various parts of the body such as face and trunk. The dermal and maxillomandibular lesions were regarded as classical manifestations of NBCCS and the patient was referred to the Dermatology department. The patient underwent total body confocal dermoscopy and suspicious lesions of the back, the left preauricular area, the right malar region and the right lumbar region were biopsied with a punch device.

Histopathological findings of the maxillomandibular cysts confirmed the diagnosis of odontogenic keratocyst and all punch biopsy specimens were reported as basal cell carcinoma (Figure 3,4). The skin lesions were resected with a healthy tissue margin of 5 millimeters and no local recurrence was seen during the follow-up period of two years. The patient was referred for further genetic tests; however, she refused the consultation.

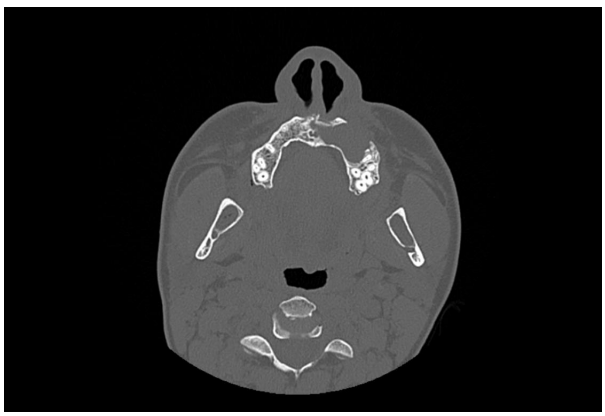

Figure 2: Preoperative computed tomography demonstrates the cystic lesion on the left side of the maxilla.

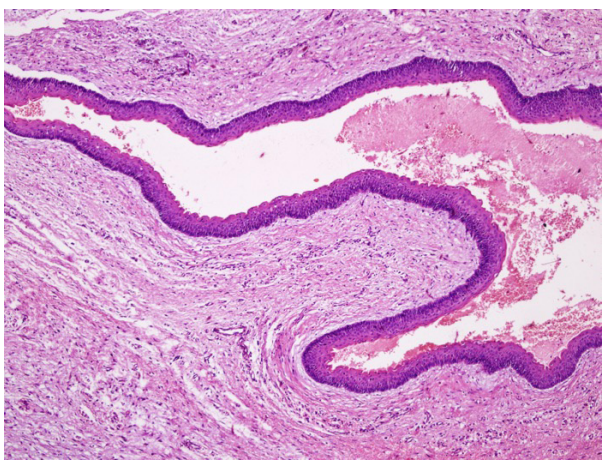

Figure 3: Histopathological appearance of the maxillary lesion showing characteristic lining of odontogenic keratocyst (OKC).

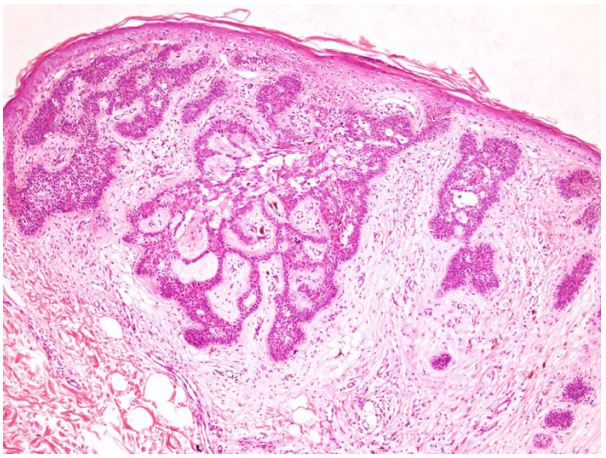

Figure 4: Histopathologic appearance of the basal cell carcinoma lesion on back of the patient.

Major criteria	Minor criteria
1. More than 2 BCCs or one under the age of 20 years 2. Odontogenic keratocysts of the jaw proven by histology 3. Three or more palmar or plantar pits 4. Bilamellar calicification of the falx cerebri 5. Bifid, fused or markedly splayed ribs 6. First degree relative of NBCC syndrome	1. Macrocephaly determined after adjustment of height 2. Congenital malformations: cleft lip or palate, frontal bossing, moderate or severe hypertelorism 3. Other skeletal abnormalities: Sprengel deformity, marked pectus deformity, marked syndactyly of the digits 4. Radiological abnormalities: Bridging of the sella turcica, vertebral anomalies, hand and foot deformities 5. Ovarian fibroma 6. Medulloblastoma

Figure 5: The diagnostic criteria of nevoid basal cell carcinoma syndrome are listed.

Biannual Plastic Reconstructive and Aesthetic Surgery and Dermatology follow-up visits were scheduled for the patient for both radiologic and dermoscopic surveillance.

DISCUSSION

Nevoid basal cell carcinoma syndrome is an uncommon autosomal dominantly inherited disorder with a mutation in the PTCH1 gene (1). Maxillomandibular cysts may be an initial sign of the syndrome (1). Patients with this syndrome may develop BCCs during childhood; however, the median age of onset is 20 years (3). In this case, the development of maxillomandibular cysts and the early onset of the dermal lesions supported the diagnosis.

In NBCCS, palmar pits occur in 75% to 90% of patients and medulloblastoma is the second most common malignancy (4).

In addition, skeletal anomalies are frequent and falx cerebri calcification, bifid ribs, frontal bossing, cleft lip/palate and vertebral fusions may be seen (3, 4). Such pathologies were not present in this case.

The diagnosis requires a multidisciplinary approach due to the variety of clinical manifestations. NBCCS may be established when two major or one major and two minor criteria are present (4). These criteria are listed in Figure 5. In this case, the patient had three major criteria including jaw cysts, multiple BCCs, palmar pitting and a minor criteria such as frontal bossing.

CONCLUSION

In NBCCS, early diagnosis enables the reduction of the complications, the prevention of the progression of the less frequent manifestations and the detection of the hereditary risk of the patient's family. Physicians should be aware of the variety of the clinical features of NBCCS.

Informed Consent: Written informed consent was obtained.

Peer Review: Externally peer-reviewed.

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