Case Report

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Gorlin Syndrome

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Abstract

Gorlin syndrome is a dominant autosomal familial disorder. The manifestations begin at an early age and a combination of phenotypic abnormalities such special facial appearance, jaw cysts and skeletal anomalies are seen in this disease. A 22-year-old woman referred to Zahedan Dental School complaining of pain on the left cheek. During the examination, several cutaneous lesions in the neck, pits in palm and sole and multiple jaw cysts were observed. According to the clinical symptoms, lesion biopsy and reports of Gorlin syndrome radiography were presented.

Introduction

Gorlin syndrome (GS) or Basal Cell Carcinoma (BCC) is a rare familial autosmal dominant disorder with full penetration and great variety. The manifestations begin at an early age and are a combination of phenotypic abnormalities including special facial appearance such as prominent forehead, flat nasal bridge, jaw cysts (odontogenic keratocyst), pits in palm and sole, skeletal and vertebral anomalies, CNS (Central Nervous System) disorders, mental retardation and multiple basal cell tumors often can be observed in this disease [1-4]. Pits in palm and sole will reveal within the second decade of life and the incidence of jaw cysts in 74% of the patients will be up to the age of 20 [3]. Most patients were white and the average age of the first tumor incidence is 17-35 years. BCC is often multiple and it usually involves the central facial regions and the upper lip [5]. In the present study, a gorlin syndrome is reported which is important in dentistry due to the incidence of multiple jaw cysts.

Introducing the patient

A 22-year-old woman referred to Zahedan School of Dentistry, the center of oral diseases complaining of pain in the left malar (cheek) region. In oral examinations and radiology images of the jaws, four third molar and canine teeth was evident. Multiple cysts were also observed beside hidden (impacted) teeth which brought about the displacement of the said teeth. Wisdom tooth (third molars) was located in the orbital floor causing pain and discomfort for the patient (Fig. 1). In the medical history of the patient, there were evidences of jaw cysts of early adolescence and the experience of jaw surgery. The patient had undergone surgery eight years ago for jaw cyst removal and histopathological lesions were reported in accordance with odontogenic keratocyst which was existed in his medical record. In examination of the patient, prominent skin lesions as papules and nodules of different sizes were evident in different regions of the neck, ear, shoulder and armpit (Fig. 2). In his history, the patient said that he has had these lesions since adolescence but they have increased in size recently. In the examination of the patient, several pits could be observed in the palm and sole preventing the patient from physical activity. Hypertelorism, flat nasal bridge and rough face were also completely evident in the patient's face.

The patient had no history of bleeding in skin lesions, anorexia, weight loss and convulsion (seizure) and also no history of abdominal pain and menstrual irregularities was reported. None of the family members had similar complaints. On chest (rib cage) radiographs, there was no evidence of bifid rib. In maxillary and mandibular CT scans, bilateral lytic lesions were observed (Fig. 2). On abdominal and pelvic ultrasound especially the uterus and ovaries, no specific lesion was reported. Brain CT scan was normal. Other tests and examinations of the patient were normal. In panoramic radiography, multiple jaw cysts were evident as several pericoronal radiolucencies

with distinct sclerotic margins associated with four impacted third molars and upper right canine (Fig. 2).

Multilocular radiolucency associated with right mandibular third molar caused the displacement of tooth toward the lower margin of the mandible. It had also made the body of ramus and coronoid notch involved. Cysts associated with right and left maxillary third molars moved the teeth toward sinus and involved all the air space of sinuses. According to the radiographies and clinical manifestations, the patient was examined more for the GS. First, the impacted third molars (wisdom teeth) were extracted under a surgery. In the histopathological examination, the tissue sample consisted of epithelial lining (6-8 cells thick), hyperchromatic basal layer and parakeratotic folded surface which in its central lumen, casein containing keratinized debris was observed and according to histopathological perspective, odontogenic keratosis was reported. Finally, the patient was referred to an oral maxillofacial specialist for surgical removal (excision) of jaw cysts.

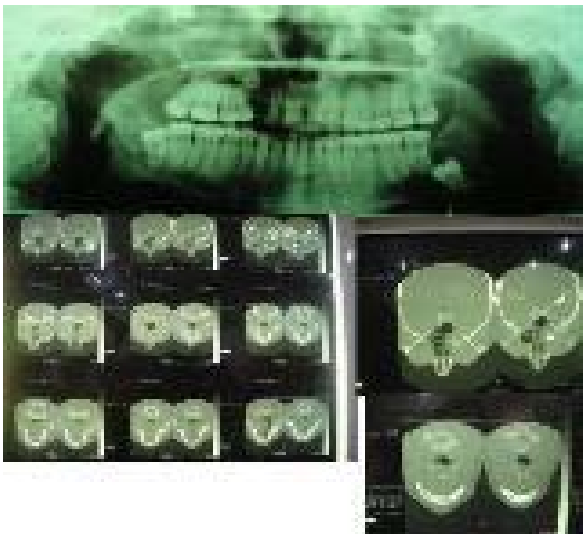

Figure 1. Position of the impacted maxillary and mandibular teeth and multiple odontogenic cysts

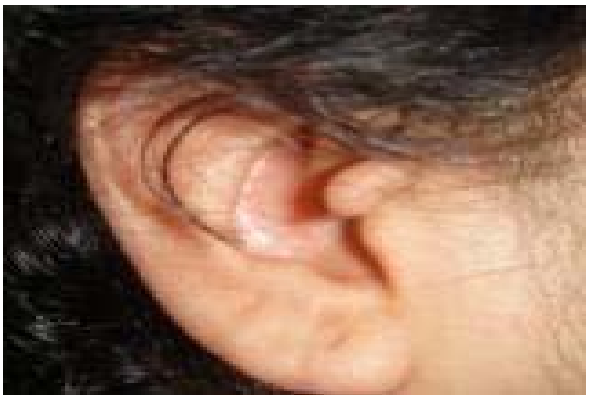

Figure 2. Cutaneous (skin) tags in the upper tragus

Discussion

Gorlin syndrome has various cutaneous and noncutaneous manifestations with different incidence [1-4]. This rare syndrome is specified with anomalies such as odontogenic keratocyst of the jaw. The prevalence of syndrome varies from 1 in 60000 to 1 in 120000 [1, 2] and the main mechanism for the incidence of syndrome has not been specified. However, mutation in the PTCH1 gene located in the long arm of chromosome 9 is responsible for the initiation of postnatal tumors. [1]Syndrome is characterized with multiple BCC, several pits in palm and sole, jaw cysts and skeletal anomalies. Jaw cysts are of early manifestations of the disease and the most common complication of the syndrome. Mandible is afflicted more than maxilla but in the present patient both of the jaws (mandible and maxilla) had been afflicted (involved). The cysts are usually asymptomatic, but they may cause pain and swelling of the jaw, especially when it causes bone erosion. This patient came while complaining of pain in the left cheek. Histologically, the cysts have odontogenic keratocyst characteristics [6]. This patient also had multiple odontogenic keratocyst cysts in the jaws. BCC usually occurs in the early youth and is multiple. These cutaneous lesions along with jaw cysts help the early diagnosis of the disease. Despite the high incidence of BCC in this syndrome, the present patient lacked this complication; however, the lesions are likely to occur up to the age of 30 [1].

Other skin manifestations are cutaneous millia and dermoid cyst. Skin (cutaneous) tags around the neck pathologically being similar to BCC but not invasive, are other manifestations of the disease [2]. Cardiac and ovarian fibromas occur in 2-20% of the cases. Cardiac fibromas are present at birth. Ovarian fibromas are discovered (detected) accidentally on ultrasound examination [7]. In this patient, no specific lesion was reported in the abdominal and pelvic ultrasound. Other manifestations of the disease are pits in the palm and sole which will reveal within the second decade of life [1-4]. This patient had also mentioned some small pitted lesions on the palm and sole from the second decade. In many cases, the presence of some symptoms such as the incidence of multiple BCC at younger ages, jaw cysts and pits on the palm and sole will make the diagnosis easy. 60% of the patients with GS have mutation in PTCH gene which is detected by sequencing. So, we can use them to detect unusual cases of illness [1].

The following diagnostic criterions have been suggested for cases when diagnosis is difficult [8]. Two major criteria or one major and two minor criteria are essential for diagnosis. Major criteria include: the existence of more than one or two BCC in under 20-year individuals, odontogenic keratocyst before the age of 15, the existence of more than 3 pits on the palm and sole, early calcification of falx celebri (before the age of 20), fusion, bifid or arched ribs, and mutation of PTHC1 gene in normal tissues. Minor criteria includes: macrochephaly, congenital malformations such as: prominent forehead, rough face, Hypertelorism and skeletal anomalies like: sprengel deformity and syndactili (deformity of fingers), and abnormalities (anomalies) which can be observed on radiologic images including: sella turcica bridge, hand

and foot lucencies, bifid rib, vertebral body fusion and ovarian fibroma.

Accordingly, the patient has two major criteria (multiple odontogenic cyst and pits on the palm and sole) and one minor criterion (flat nose, rough face and hypertelorism). So, he was treated with definite diagnosis of Gorlin syndrome. The most important points in controlling GS are repeated examinations, protection against sun and immediate surgical and non surgical treatment of small tumors. There was no difference in the longevity of patients with GS with healthy individuals. The main problem of these patients is the problem of beauty which occurs after the treatment of BCC lesions and with fewer degrees of jaw cyst surgery [1]. Early diagnosis and immediate treatment of the disease would be a great help for decreasing the complications of the disease and improving the personal quality of life.

Authors' Contributions

All authors had equal role in design, work, statistical analysis and manuscript writing.

Conflict of Interest

The authors declare no conflict of interest.

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