Goltz-Gorlin Syndrome: Report of Two Cases

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Abstract

Goltz-Gorlin syndrome or Nevoid Basal Cell Carcinoma syndrome (NBCCS) is an infrequent multisystemic disease that is inherited in a dominant autosomal pattern, which shows a high level of penetrance and variable expressiveness. The syndrome is associated with multiple Keratocystic Odontogenic Tumors (KCOT) in 90% of cases. In addition to jaw cysts, multiple basal cell carcinomas and skeletal anomalies are common findings. KCOTs associated with this syndrome can be seen in both maxilla and mandible but there is more tendencies to posterior mandible and the recurrence rate is more than nonsyndromic cases.

In this article we describe two patients with diagnosis of Goltz-Gorlin syndrome, its typical findings and finally we discuss about the treatment method used for jaw cysts and the outcome of 1 year follow-up.

Keywords: Goltz-Gorlin syndrome; Nevoid basal cell carcinoma; Keratocystic odontogenic tumors

Introduction

Goltz-Gorlin syndrome or Nevoid Basal Cell Carcinoma syndrome (NBCCS) is an infrequent multisystemic disease that is inherited in a dominant autosomal pattern, which shows a high level of penetrance and variable expressiveness. In 1894, Jarisch and White made the first descriptions of patients with this syndrome, highlighting the presence of multiple basal cell carcinomas. Then in 1960, Robert W Goltz and Robert J Gorlin described a triad of signs of this disease including; multiple nevoid basal cell carcinomas, multiple jaw keratocysts and bifid ribs [1,2]. Pathogenesis of the syndrome is attributed to abnormalities in the long arm of chromosome 9 (q22.3-q31) and loss of, or mutations of human patched gene (PTCH1 gene) [2].

The syndrome is associated with multiple Keratocystic Odontogenic Tumors (KCOT) in 90% of cases. In addition to jaw cysts, multiple basal cell carcinomas and skeletal anomalies are common findings. Skeletal findings are; bifid ribs, vertebral anomalies, bossing of frontal and temporoparietal region, hypertelorism, palmar and plantar pit and fissures, Calcification of falx cerebri, ocular and central nervous system lesions, cleft lip and palate, mandibular prognathism and in some rare cases the ovarian fibroma [3]. KCOTs associated with this syndrome can be seen in both maxilla and mandible but there is more tendencies to posterior mandible [4] and the recurrence rate is more than nonsyndromic cases [5]. In this article we describe two patients with diagnosis of Goltz-Gorlin syndrome, typical findings of it and finally we discuss about the treatment method used for jaw cysts and the outcome of 1 year follow-up.

Case Presentation

Case 1

A 15 years old boy by a chief complain of pain in dental region referred to dentist and an Orthopantomogram (OPG) asked for him. In the OPG of patient the multiple jaw lesions has been diagnosed and patient has been referred to Mashhad dental faculty. In clinical examination, the patient had a slight frontal and temporal bossing. There were no hypertelorism but he had unibrow. There was no pigmented lesion in any region of patient’s body (Figure 1).

In OPG (Figure 2), radiolucent lesions related to bilateral mandibular and right maxillary impacted third molars were observed. In the left maxilla second and third molars were impacted.

In the right mandible, radiolucent lesion with well defined cortical border and large extension from sigmoid notch to apical region of distal root of right 1st molar was observed that led to displacement of impacted third molar to the gonial angle. Also in the left side of mandible there was a well defined radiolucent lesion with cortical border in relation to impacted third
molar that was 14*7mm dimensions. There was a slight tooth bud displacement in this region. And in the right side of maxilla there was a radiolucent, well defined with cortical border lesion related impacted third molar that was caused the displacement of the tooth. In the skull radiography, calcification of Falx cerebri was observed in form of radiopaque lines in the center of the skull which is a typical view of this syndrome (Figure 3).

Histopathological evaluation of jaw lesions revealed cystic wall with ulcerated areas, squamous epithelium and wavy parakeratin. According to these findings, the diagnosis was Keratocystic odontogenic tumor (KCOT).

Treatment plan for the patient was including the prophylactic removal of impacted left maxillary teeth, removal of impacted third molars with enucleation of related KCOT in the left mandibular and right maxillary region and marsupialization of the right mandibular cyst. Marsupialization was done by creating a window in the epithelium of cyst and suturing it to the surrounding periosite and then to keep it open by an acrylic obturator with a process extended to the cyst was made. Patient followed every month and at third month, an OPG was taken that revealed calcification of surgical site in left side of mandible and decompression of cyst followed by upward movement of impacted third molar in right side of mandible (Figure 4). No new lesions seen on the provided image.

Case 2

A 21 years old man with chief complains of pain and swelling of lower jaw came and an OPG was ordered. In his OPG there were multiple radiolucent lesions in both mandible and maxilla (Figure 5). In clinical examination, the patient had a slight frontal bossing but there was no hypertelorism. Also he had pigmented lesions in multiple sites of his body. He had unibrow too (Figure 6). In the panoramic and cone-beam computed tomography (Figure 7), there were radiolucent lesions in 3 sites of jaws; right ramus and retromolar of mandible, left retromolar of the mandible and distal of right maxillary 2nd molar. There were no impacted teeth and the patient had no third molars. In the right side of mandible, the radiolucent lesion was in the distal of 2nd molar and posterior extension to mandibular ramus. The lesion had a shape like clover leaves with well defined and cortical border and approximate dimensions of 30*10mm that was caused bone
perforation in retromolar region. In the left side of mandible, there was a radiolucent lesion in the distal of 2nd molar with well-defined and cortical border and approximate dimension of 16*20mm. This lesion was extended posterior to the anterior of ramus. In the right maxillary, a radiolucent lesion in the distal of 2nd molar with well-defined and cortical border and approximate dimension of 28*25mm. This lesion was extended anteroposteriorly and in the anterior extension, was invaded into the maxillary sinus. Also, buccal cortex of maxilla in distal of 2nd molar was destroyed completely. PA skull radiography was obtained in which the calcification of falx cerebri was observed (Figure 8) and in the chest radiography, there was no bifid rib (Figure 9).

Surgery of jaw lesions were performed under general anesthesia. In maxilla the radiolucent lesion enucleated completely and due to involvement of surrounding bone, the 2nd molar extracted too. In mandible, the treatment plan was to marsupialization of cystic lesion in both sides, so a window in the epithelium of cystic lesions was made and kept open by using an acrylic obturator in both sides. Histopathologic diagnosis of all three regions was KCOT.

Also we did a biopsy from skin lesion of facial region and the histopathologic diagnosis of it was Basal Cell Carcinoma (BCC). The patient followed up monthly and OPG taken at 3rd month after surgery showed the calcification in right maxillary region and reduce in size of mandibular lesions. The patient referred to a dermatologist for treatment of skin lesions.

Discussion

The Gorlin syndrome has diagnostic features such as multiple jaw KCOTs, NBCC, calcification of falx cerebri, palmar and plantar pit and fissure, bifid ribs, hypertelorism, frontal and temporal bossing and etc [6] there were KCOT in two side of mandible in our first case and two side of mandible and right side of maxilla in the second case and also there was calcification of falx cerebri in both cases. These findings were similar to the diagnostic findings of Kavoosi and colleagues [7]. According to Habibi and colleagues average age of diagnosis of Gorlin syndrome in Iranian population is 35/12 years [8], which in our cases it was lower (15 years old in case 1 and 21 years old in case 2). Multiple KCOTs is the primary diagnostic symptom of this syndrome, as in our both cases.

According to Habibi and colleagues, there were multiple KCOTs in all of 19 cases they studied [8] according to Gonzalez et al. [9]. These cysts are more willing to engage mandible. Which It was observed in our patients too (Figure 1 and 6). According to Habibi et al [8], the calcification of falx cerebri exists in 89% of Iranian population suffering from this syndrome which was observed in our both patients. Multiple Basal cell carcinoma usually appears in puberty or the second and third decades of life [9], in our first case there was no skin lesion but the second patient had multiple skin lesions on his face and upper limb that biopsy from facial lesions revealed BCCs. KCOT treatment depends on several factors, including age, size and location and extension of lesion, perforation of cortical
bone and invasion to soft tissue [6,10]. According to this, treatments are generally classified as conservative or aggressive; Conservative treatment generally includes simple enucleation- with or without curettage, or marsupialization. Aggressive treatment generally includes peripheral ostectomy, chemical curettage with Carnoy’s solution, cryotherapy, or electrocautery and resection [10]. Different recurrence rates have been reported with each of these methods. According to study of Kaczmaryc et al. [11] on the choice of treatment method and recurrence rate of jaw lesions, the lowest recurrence rate was in resection and the highest rate was in enucleation (with or without adjunct therapy). Also, according to Titinchi and colleagues, recurrence rate of KCOT is reported 29/2% that in their study, the highest recurrence was associated with marsupialization [12]. In our first patient that we did enucleation in right maxilla and left mandible and marsupialization in right side of mandible for treatment of KCOT, in 10 month follow-up period, there was no recurrence and the size of right mandibular lesion was reduced.

In 18 months follow-up period of our second patient, there was no recurrence in maxilla which was treated by enucleation. In the mandibular lesions, according to OPG radiographies taken 3,6 and 12 months after surgery, the lesions was progressing towards healing but in OPG taken at month 18, the radiolucency was a little increased. So we requested a CBCT from both side of mandible and according to it, lesion’s size was reduced but the cysts needed enucleation that was done by a second surgery.

**Conclusion**

Gorlin-Goltz syndrome is a rare autosomal dominant disorder that involves multiple organ systems and if it be diagnosed as early as possible, there is the possibility of more effective treatment and prevention of some findings. So in suspected cases of this syndrome, Counseling and referral to other related specialties for a more comprehensive diagnostic evaluation is recommended.

**References**