TREATMENT OF ODONTOGENIC KEROTOCYSTS IN NEVOID BASAL-CELL CARCINOMA SYNDROME – CASE REPORT

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Abstract

Background: Gorlin-Goltz syndrome is a rare disease with different diagnostic characteristics such as multiple odontogenic keratocytes of the jaws, basal cell carcinoma of the skin, palmar and plantar pits, frontal bossing, hypertelorism, and falx cerebri calcification. Herein, we present a rare case of Gorlin-Goltz syndrome.

Case report: A 38-year-old woman was referred to our specialized dentistry clinic affiliated to Tehran University of Medical Sciences with the chief complaint of bilateral mandibular swelling without pain and parasthesia. In intraoral clinical examination, bilateral inflammation in the buccal vestibule of the mandible was observed which had bone consistency on palpation but without any tenderness.

In the panoramic view a radiolucent lesion with defined cortical borders extending from the first tooth on the left to the distal root of the 6th lower tooth, presenting a scallop pattern between the roots, was revealed in the right side. To treat the jaw cysts an incisional biopsy was initially performed and after confirmation of odontogenic keratocytes and regarding the multiple and wide spread lesions, decompression was done on the left side to treat the cysts of the mandibular body.

Conclusion: Early diagnosis and treatment of Gorlin-Goltz syndrome is of great importance in preventing long-term complications such as malignancy, destruction and malformation of the jaws.

Kerwords: Decompression, Falx cerebri calcification, Gorlin-Goltz syndrome, Odontogenic keratocystes.

Introduction

The Gorlin-Goltz syndrome was initially reported by Jarisch in 1894.[1] Binkley and Johnson once again defined it in 1951. In 1960 Gorlin *et al*. extracted it from the scientific archives, classified and delineated it.[2] This syndrome has been mentioned in different studies with various terms such as "Basal cell nevus - Bifid rib syndrome" and "Hereditary Cutaneomandibular Polyoncosis".[3-5]

Gorlin-Goltz syndrome or Basal Cell Nevus Syndrome (BCNS) is one of the rare hereditary autosomal dominant disorders which has variable manifestations.[6] Its prevalence ranges from 1 in 57000 to 1 in 256000 individuals and its incidence is similar in both genders.[5] A mutation in a tumor suppressor gene (PTCH1 gene) residing on the long arm of chromosome 9 is responsible for the formation of many postnatal tumors in this disease.[6,7] Such mutations have been detected in 60-85% of individuals tested by sequencing of PTCH1.[8] The patients' age varies between 6 to 78 yrs at diagnosis and the peak prevalence has been reported in the 3rd decade of life with a mean age of 32.8 years. Around 70.5% of lesions in the jaw are seen in the mandible and 6% of the cases accompanied by basal cell have the main signs of this syndrome such as odontogenic carcinoma.[9] The clinical manifestations of this syndrome are as follows, respectively:

1. Skin and soft tissue anomalies including basal cell nevus in the lumbar and flank regions, basal cell carcinoma, palmar and plantar keratosis, skin calcinosis and superficial fibromatosis
2. Tooth and jaw anomalies consisting of multiple odontogenic keratocytes in the jaws besides oligodontia
3. Bone anomalies which are present in 75% of the cases and include fusion, spino bifidity, multiple bifid ribs of the cervical vertebrae, bridging of the sella turcica, shoulder deformity, shortening of the 4th metacarpal and metatarsal bones and polydactyly
4. Neurologic anomalies including mental retardation, extracorporeal calcification, falx cerebri and tentorium calcification, non-formation of corpus callosum and congenital hydrocephaly
5. Eye disorders including congenital cataract, sexual dysfunction and hypogonadism in males and ovarian tumors in females.[5,10-14]

The laboratory abnormalities of this syndrome include:

1. Dysfunction in the diuresis of phosphor in response to frequent injections of the parathyroid hormone (Elsorth-Hward test).
2. Calcium retention following the calcium load test.
3. Pseudohyperparathyroidism in some patients.[5-15]
4. The occurrence of odontogenic tumors of keratocytes is usually the first sign of this syndrome.[12]

The main considerable point in these patients is the incidence of odontogenic keratocytes and their recurrence in the following months and years in the areas which were previously free of cysts.[7] Odontogenic keratocysts are intraosseous lesions of the jaw and present in the form of a single or multiple cavities with potential for local destruction and tendency for multiplicity which is especially true in Gorlin-Goltz syndrome.[13,14] This lesion is commonly symptom-free but may occasionally present with diffuse pain.

In most cases the radiographic presentation of the lesions is changes in the follicular space size and shape which can be observed by CT scan or simple x-ray imaging.[9]

In the x-ray view, the cysts may present as dentigerous cysts.[6] In general, the cystic lesions in the jaws in BCNS

have been described as multiple, distinct radiolucencies with well-defined borders.[6]
Three surgical methods have been proposed for removing theses cysts as follows:

1. Enucleation and curettage and initial closing of the lesion
2. Marsupialization and leaving the cavity open
3. Enucleation

No significant difference in the recurrence rate has been mentioned between theses three methods[16] and the entity of OKCs is related to the genetically high recurrence rate in this syndrome.[17]

Case report

A 38-year-old woman was referred to the dental clinic of Tehran University of Medical Sciences with bilateral swelling of the mandible without any pain and paresthesia. She had been suffering from this condition since 6 months before. She had basic education and was forced to leave school due her average IQ. There was no family history of jaw abnormalities. On intraoral clinical examination she had bilateral swelling in the buccal vestibules with no tenderness. The color of the mucus was normal but dental displacement and changes inocclusion were evident. Her teeth had no mobility. [Figure 1]

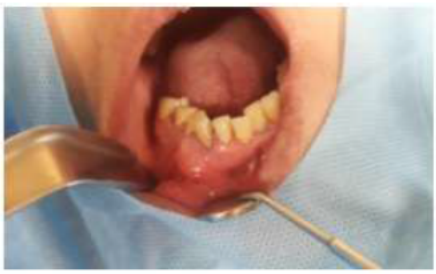

Figure 1: Before surgery; dental displacement and inflammation

The involved teeth in the right side had previously undergone endodontic therapy. Moreover, all her teeth in the left side had been extracted following the initial stages of therapy.

In the oral panoramic radiograph, a radiolucent lesion with defined cortical borders was observed in the right side extending from the 1st tooth on the left to the distal root of the 6th lower tooth, creating a scallop pattern between the roots. [Figure 2]

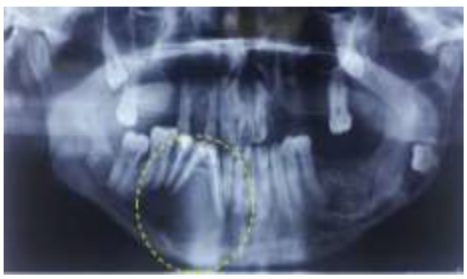

Figure 2: Dental panoramic radiography before treatment

The lesions had caused displacement of the involved teeth and displaced the lower alveolar canal downwards. The upper canal border was not observed adjacent to the lesion. In addition, another lesion had extended from the distal part of the 7th tooth in the right lower side to the lower part of the ramus with defined cortical borders. On the left side, two separate lucent unilocular lesions with defined cortical borders were present, one extending from the distal part of the 5th left lower tooth to the lower part of the ramus; involvement of the pericronal region of the 8th left-sided tooth was impacted and extended to the sigmoid notch. In the right side of the upper jaw, a lucent lesion was seen with non-defined borders extending from the distal part of the 2nd upper right tooth to the 4th upper right tooth. In the left side, two radiolucent lesions with defined cortical borders, one extending from midline to the 2nd upper left tooth and the impacted pericoronal involvement of the 8th left tooth were apparent. [Figure 3]

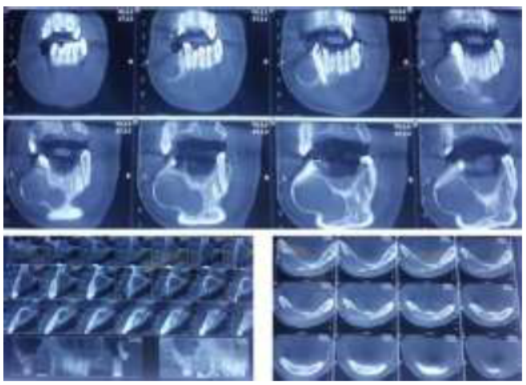

Figure 3: Cone beam computed tomography (CBCT) of the upper jaw(a), and lower jaw(b=Axial View),(c=Coronal View) before treatment.

In the anterior-posterior skull view, calcification of the falx cerebri was observed [Figure 4] The central nervous system and cardiovascular systems were both normal. Her skull circumference had also increased to 58 cm (normal size: 55 cm).

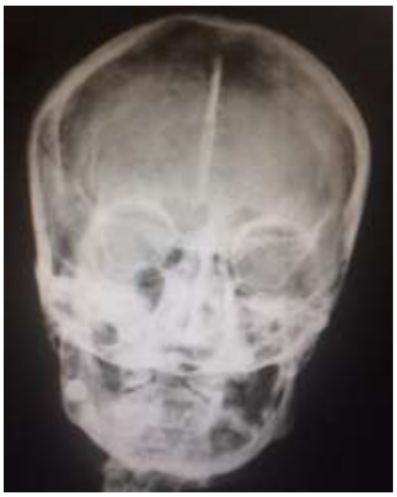

Figure 4: Calcification of the falx cerebri in X-ray imaging.

Regarding the evaluation of ovarian cysts, she was referred to a gynecologist. A single cyst sized 33mm in diameter and a follicle sized 28 mm in diameter weres revealed in her left ovary. A small amount of free fluid was reported in the cul-de-sac while the right ovary was normal. [Figure 5]

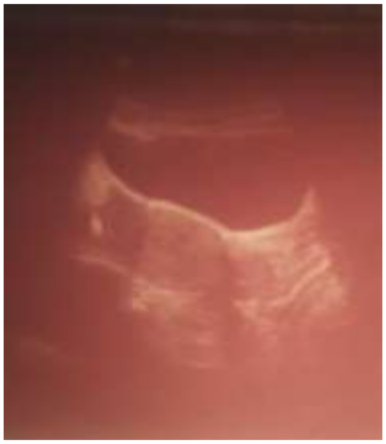

Figure 5: Ultrasound imaging of the left ovary.

The most prominant mole in the patient's face was biopsied [Figure 6] revealing basal cell carcinoma of the adenoid type.

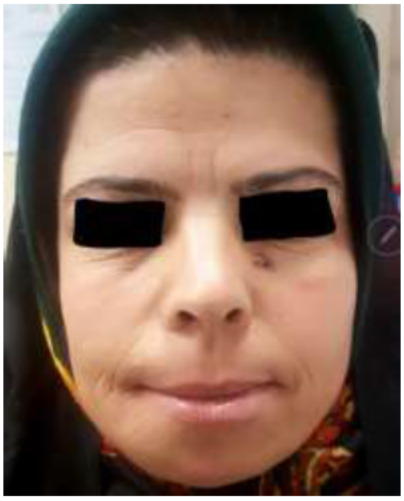

Figure 6: The biggest nevus in upper face.

Thus, she was referred to a dermatologist for further clinical treatment and follow up. In addition, multiple moles were visible on the patient's body. [Figure 7]

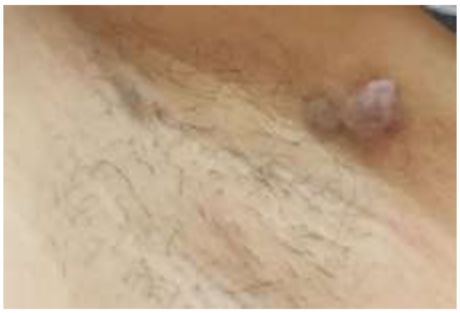

Figure 7: Nevus on the axillary.

In order to treat the cysts in the jaw, an incisional biopsy was initially performed keratin withdrawal was evident during the operasion; [Figure 8]

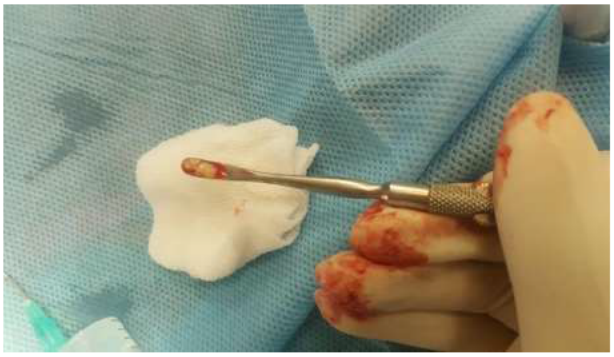

Figure 8: Keratin secretion during biopsy of the lesion.

After confirmation of the ortho keratocyst and given the multiple wide-spread skin lesions, marsupialization was performed on the left side to treat the cysts of the mandible's body. [Figure 9]

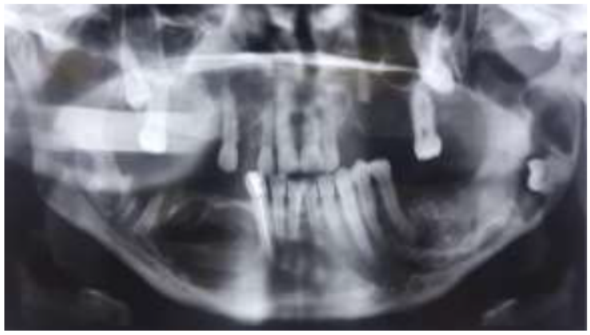

Figure 6: The radiographic view after the first marsupialization treatment on the right and left side.

After the 6-month follow up and bone formation in the right side, decompression was done on the body and the 4th, 5th, 6th and 7th teeth were extracted. A nelaton with a 4mm diameter was placed in that site as a valve to relieve pressure.

The patient was followed up for 4 months revealing a gradual reduction in the cyst's size. In the next step, all jaw cysts were removed by the enucleation surgical technique and curettage under general anesthesia.

The patient was anesthetized with nasal intubation through the left passage; the pharyngeal pack was placed and with an intraoral incision in the anterior ridge of the myelohyoid of the left mandible side it was extended to the ridge of the mandible, the flap was elevated and the lesion was accessed by creating a valve in the ramus region. The lesion was enucleated and the impacted wisdom tooth on the same side was extracted. A 3mm margin around the lesion was freezed and curettaged with a round bur. The lesion was then accessed in the right body region through the mentioned valve and was enucleated by splitting into two pieces. [Figure 10]

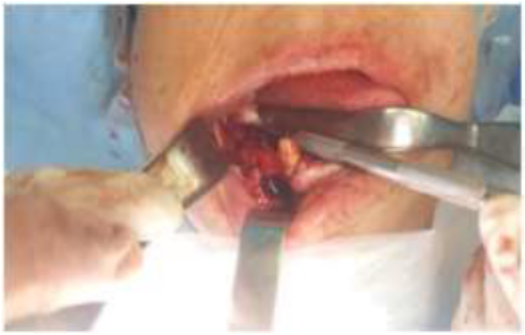

Figure 10: Surgery in lower jaw

In the upper jaw, with a crestal incision in the anterior maxilla, the flap was elevated and the lesion was approached through the buccal area on the right and the palatal area on the left. [Figure 11]

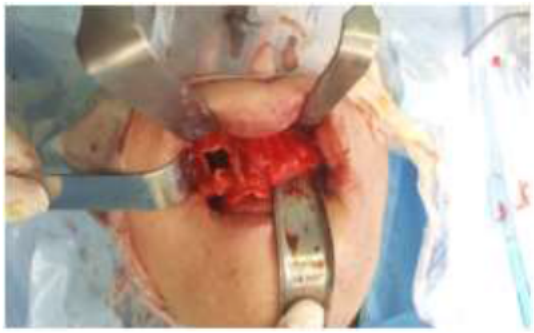

Figure 11: Surgery in upper jaw

These areas were also curettaged with round bur, similar to the lower jaw.

The bilateral upper wisdom teeth were extracted with a triangular incision in the posterior part of the jaws.
The crestal incision was made on the patient's ridge; the cyst surrounding the upper left wisdom tooth involving the maxillary sinus on the same side was extracted and the region was curettaged. These areas were washed and the bone irregularities were smoothened by a rongeur and the cuts were sutured by 3-0 Vicryl sutures. The patient's pharyngeal pack was then removed and a compression bandage was placed on the patient's face. [Figure 12]

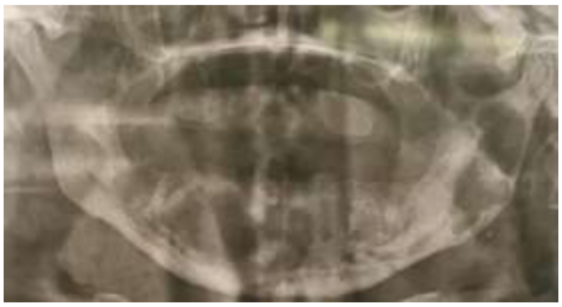

Figure 12: Radiographic view at the end of the surgical procedure

The removed tissues were sent to the lab for histopathological examination. In the lab report, OKC was seen in the left mandibular ramus, and radicular cyst was reported in the mandibular body. Moreover, keratocysts were reported on the right side of the mandible and on both sides of the maxilla.

The patient was advised to visit the clinic on a weekly basis in the first month and then every 3, 6 and 12 month for radiologic and clinical examination

Discussion

Gorlin-Goltz syndrome is of great importance for dentists and surgeons as the appearance of keratocytes with a dental origin may be the initial sign of this syndrome; occurring in 74-90% of such cases.

OKCs have a high recurrence rate varying in number from 1 to 28. The treatment of choice is surgery with the aim of complete resection.[5,18] The presentation of BCC in such patients may vary from small papules to large lesions ranging in number from a few to a few thousand. In limited numbers, surgery can be performed whereas in greater numbers nonsurgical approaches along with follow up are the treatment of choice. Radiotherapy should be avoided in such patients. The use of vitamin A analogues and avoidance of long-term sun exposure can be helpful in preventing the occurrence of new BCC lesions.[5,11]

Different studies have reported various types of Gorlin-Goltz syndrome. Shahsavan *et al*. in 2009 reported a case of a 23-year-old man with Gorlin-Goltz syndrome whom was diagnosed due to multiple, recurrent and in some cases infectious OKCs. He also had polydactyly in his left hand and foot, palmar and platar keratosis, frontal bone prominence, widened nasal saddle, hypertelorism, multiple bifid ribs, Falx cerebri calcification, BCNS nf the left lumbar side and the congenital absence of all four wisdom teeth; he had not been previously diagnosed with this disease.[19]

Kavoosi *et al*. in 2012 reported another case of Gorlin-Goltz syndrome. The patient was a 23-year-old man whom had pus secretion from the left side of the lower jaw (buccal or first molar) was referred to the radiology unit of the Dentistry School. In the panoramic radiographic view, multiple cystic lesions in the mandible and impacted teeth in the maxilla were diagnosed. In the microscopic view, epithelium of the odontogenic cyst, keratin debries and palisaded basal cells were observed revealing the diagnosis of multiple odontogenic keratocytes.

Treatment was initially done by the marsupialization technique followed by enucleation and curettage. No recurrence was reported in the 3-month follow up. They concluded that the early diagnosis of BCNS by a dentist or dental specialists has a significant role in the patient's prognosis.[20]

In the different studies having reported this syndrome including the study by Kimonis *et al*[21] in 1997 on 105 patients during 12 years, Muzio *et al*[5] study in 1999 on 37 patients and Friedrich study[19] in 2007 on 17, keratocytes of the jaws, falx cerebri calcification and basal cell carcinoma

were mentioned as the main features of Gorlin-Goltz syndrome. Palmar and plantar hyperkeratosis, skeletal abnormalities and facial deformities are other manifestations of this syndrome which have a lower prevalence. Moreover, in 5% of the cases mild intellectual deficit and malignant brain tumors (mostly as meduloamyloblastoma) have been observed.[22]

Taken together, the early diagnosis of Gorlin-Goltz syndrome by the dentist or dental experts has a significant role in the patient's prognosis.

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