Management of multiple odontogenic keratocyst in a case of Gorlin-Goltz syndrome and literature review

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Abstract

Gorlin-Goltz syndrome represents a series of multiorgan abnormalities known to be the consequence of abnormalities in the human patched gene and increases the risk of developing various cancerous and non-cancerous tumors. It is a rare autosomal dominant syndrome with a high level of penetrance and variable expressiveness. We present a case of Gorlin-Goltz syndrome with no familial history, who was incidentally diagnosed with syndrome and presenting with multiple odontogenic keratocysts, palmar pits, bilamellar calcification of falx cerebri, and bifid ribs. This case highlights the importance of early diagnosis, the need for awareness of diagnostic criteria in cases with no typical skin lesions followed by multidisciplinary approach to delay the progress of the syndrome. Thus, it is important that different health specialists be aware of the main features of this syndrome so as to diagnose the condition at earliest to provide appropriate surgical treatment.

Keywords: Gorlin-Goltz syndrome, odontogenic keratocysts, PTCH1 gene

Introduction

Gorlin-Goltz syndrome a rare multisystemic disease which is transmitted as an autosomal dominant triad with high penetrance and variable expressivity and estimated prevalence varying from 1/57,000 to 1/256,000 people. It affects male and female equally, with involvement of the first, second, and third decade of life. It is associated with different signs and symptoms causing skin, skeletal, craniofacial, neurological, oropharyngeal, genitourinary, and cardiac abnormalities.¹,²

This syndrome has received several names such as “basal-cell nevus syndrome,” “nevoid basal-cell carcinoma syndrome” ever since Jarisch and White made the first description of the syndrome in 1894. In 1960, Dr. Robert Gorlin and Dr. Robert Goltz established a classical triad: Multiple basocellular epitheliomas, keratocysts in the jaw, and bifid ribs for diagnosis of this syndrome.²

Case Report

We describe a case of a 19-year-old male who reported to our unit for the replacement of missing teeth in the upper left front region, incidentally presented with multiple odontogenic cyst and was diagnosed with Gorlin-Goltz syndrome. It was unusual that there were multiple missing permanent teeth for his age. There was no relevant history pertaining to underlying systemic illness. On general examination, he presented with normal intellectual ability but had minor skeletal anomalies such as drooping shoulder, frontal bossing, and palmar pits. Orthopantomogram revealed multiple impacted teeth with associated odontogenic cyst involving maxilla and mandible in all the quadrants. Posteroanterior (PA) skull showed the presence of calcification of falx cerebri and bifid ribs. This case highlights the importance of early diagnosis, the need for awareness of diagnostic criteria in cases with no typical skin lesions followed by multidisciplinary approach to delay the progress of the syndrome. Thus, it is important that different health specialists be aware of the main features of this syndrome so as to diagnose the condition at earliest to provide appropriate surgical treatment.
Figure 4. The lining is separated with the help of periosteal elevator and is gently eased away from the cavity wall. After complete enucleation of the cyst along with the impacted tooth, sterile gauze swab presoaked with Carnoy’s solution was placed in the lumen of the defect and left there for 3 min protecting the adjacent soft tissue with vaseline gauze; followed by irrigation with saline to visualize the remnants facilitating its easy removal. Finally, flaps closed with 3-0 vicryl sutures. Similar procedure carried out on the left side of the mandible, bilateral maxillary third molar region, and right maxillary canine region and cystic lesions which were enucleated sent for histopathological examination [Figure 5]. Microscopic examination suggested of parakeratinized odontogenic epithelium 6–8 cell thick with keratin flakes in the luminal portion as well as interface between epithelium and connective tissue is flat, and thus histologically diagnosed to be an odontogenic keratocyst-parakeratinized variant. Thus, the final diagnosis of Gorlin-Goltz syndrome made based on the clinical, radiographic, and histopathological finding. There was no post-operative complication and patient was followed up periodically for 1 year with no evidence of recurrence.

Discussion

Gorlin-Goltz syndrome has a variable spectrum of presentation but principally presented as a triad of multiple basal cell nevi, jaw keratocysts, and skeletal anomalies. Pathogenesis is attributed to loss of human patched gene (PTCH1 gene) which is a tumor suppressor gene linked to the long arm of chromosome 9 (q22.3-q31). The diagnosis is based on the 1991 Evans et al. diagnostic criteria which were later modified by Kimonis et al. in 1997 and Bree et al. in 2011 [Table 1]. Diagnosis is made with the presence of two major criteria, 1 major criteria and molecular confirmation or 1 major and 2 minor criteria.2-5

The age predilection for this condition is in the 1–3rd decade of life supporting our case who presented to us in the second decade of life. Although in our case, the patient did not present with basal cell carcinoma, there were other major and minor criteria fulfilling the diagnosis of the syndrome. Three major criteria: Histologically proven odontogenic keratocyst of jaw, palmar pits, and calcification of falx cerebri; minor criteria, i.e., skeletal anomalies such as frontal bossing, bifid ribs, mild
Macrocephaly, and oropharyngeal anomalies such as impacted teeth and high arched palate.

Multiple odontogenic keratocysts are the first as well as the most consistent and representative sign in this syndrome with an incidence of 75% and mandible being the most common site.\(^3\)\(^-\)\(^7\) There may be remarkably few symptoms until cyst reach a large size, especially when in the ascending ramus which supports our case who was asymptomatic at presentation and multiple cystic lesions were noted on routine radiographic examination. Microscopically, they are multilocular with a parakeratinized stratified squamous epithelium most common type (96%) which is identical to our case.\(^5\)

Management of odontogenic keratocyst is controversial due to the high recurrence rate as stated by Forsell et al. which is 63% in keratocysts associated to the syndrome due to the inherited tendency of the basal epithelium to develop new cysts and 37% in the isolated ones.\(^1\)\(^-\)\(^7\) The therapeutic techniques vary from simple enucleation with curettage to enucleation with peripheral ostectomy or to osseous resection in block along with cryotherapy or Carnoy’s solution.\(^6\)\(^-\)\(^8\) In this case, surgical enucleation of all the cystic lesions with chemical cauterization was done.

**Conclusion**

This case highlights the importance of Gorlin-Goltz syndrome which may be undiagnosed, especially in young people without any skin lesion. Thus, it is important that different health specialists be aware of the main features of this syndrome so as to diagnose the condition at earliest to provide appropriate surgical treatment.

**Clinical Significance**

Gorlin-Goltz syndrome an uncommon multisystemic disease, which may be underdiagnosed with a high morbidity. Early diagnosis and treatment, family screening, and genetic counseling are essential due to its association with aggressive basal cell carcinomas and malignant neoplasias. Thus, patients are offered a multidisciplinary lifelong surveillance program to minimize the skeletal deformities and to reduce the lethal complications as early as in childhood.

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