

CASE REPORT

MULTIPLE KERATOCYSTIC ODONTOGENIC TUMORS: A PRESENTATION OF GORLIN GOLTZ SYNDROME

Selven Thirumalai¹, Sumir Gandhi², Inderjot Singh³, Navpreet Bedi⁴

¹⁻⁴Department of Oral & Maxillofacial Surgery, Christian Dental College, Brown Road, Ludhiana.


ABSTRACT:

Background :Gorlin Goltz syndrome is a rare inherited disorder of multiple Keratocystic odontogenic tumors (KCOT) with cutaneous, dento-osseous, ophthalmic, neurological abnormalities. Characteristic findings include multiple KCOTs and basal cell nevi. The KCOT should be treated aggressively due to its recurrence and complications. **Method** :We discuss two cases of multiple KCOT in young patients, who presented with primary complaints related to the presence of keratocystic odontogenic tumor and were diagnosed as Gorlin Goltz syndrome based on the presence of major and minor diagnostic criteria. **Results** :Both our cases presented with complaints related to the presence of KCOTs which were treated surgically without evidence of recurrence till date. **Conclusions** :In Gorlin Goltz syndrome, KCOT's are often the first clinical feature to be recognised and could be the only condition for which the patient may seek treatment. It is therefore important that OMF surgeons have knowledge of the main features of this syndrome and a high index of suspicion be present when a patient of multiple KCOT presents to our clinics.

Keywords : Keratocystic odontogenic tumor, KCOT, Gorlin Goltz Syndrome, Odontogenic keratocyst, NBCCS.

Corresponding Author: Dr Selven Thirumalai, PG resident, Department of Oral & Maxillofacial Surgery, Christian Dental College, Brown Road, Ludhiana, Punjab

This article may be cited as: Thirumalai S, Gandhi S, Singh I, Bedi N. Multiple keratocystic odontogenic tumors: A presentation of gorlin goltz syndrome. J Adv Med Dent Scie Res 2017;5(6):1-4.

Access this article online	
Quick Response Code 	Website: www.jamdsr.com
	DOI: 10.21276/jamdsr.2017.5.6.01

INTRODUCTION

Gorlin Goltz syndrome is a rare autosomal dominant disorder characterized by multiple odontogenic keratocysts along with various cutaneous, dental, osseous, ophthalmic, neurological, and sex organ abnormalities. The most common findings include multiple odontogenic keratocysts and basal cell nevus on the skin that have an early age onset. The odontogenic keratocysts should be treated aggressively due to its potential to recur and the possibility of complications.

We present two cases of multiple odontogenic keratocysts in young patients, who were diagnosed as Gorlin Goltz syndrome based on the presence of major and minor diagnostic criteria.

Case 1:

A 23 year old gentleman presented with pain in the mandibular anterior region for the past 6 months. The pain was spontaneous, localized, mild, non-radiating, with no aggravating/relieving factors and no diurnal/positional variations. He also complained of pus discharge from the same region for the last 3 months. Examination revealed no obvious facial asymmetry/swelling. There was mild tenderness on palpation in the submental region but the mandibular lower border was uniform.

Intraoral examination revealed missing # 33,34, RCT # 32, Mobility Grade I # 31,42,41, tenderness on percussion of # 31,32. Pus Discharge was observed from the gingival sulci of left canine and premolar with expansion of buccal and lingual cortices at the same region causing vestibular obliteration and mild vestibular tenderness. The lesion was hard in consistency with a dimension of approximately 2.5 x 2 x 2 cm. Aspiration yielded 2-3 cc of pus.

Radiography revealed well defined unilocular cystic lesion at the left body and parasymphysis region, left mandibular 3rd molar region, right mandibular ramus, right maxillary canine region, right maxillary 3rd molar region, left maxillary canine region and impacted # 13, 18, 38.

A provisional diagnosis of infected KCOT was made, with a differential diagnosis of dentigerous cyst. Because of presence of multiple cystic lesions a differential diagnosis of Gorlin Goltz syndrome was proposed. Further examination revealed multiple nevi on the face, shoulders, chest and back, palmar pits, plantar hyperkeratosis, pectus excavatum and a bifid 4th rib. (Fig I)

Enucleation of the lesions was performed under general anesthesia. Impacted teeth were extracted and Functional endoscopic sinus surgery (FESS) with middle meatus antrostomy was done.

Histopathological study showed parakeratinized cystic lining that was 6-10 layers with areas of atrophy and hyperplasia thick with underlying fibrovascular connective tissue capsule. Basal cells were tall columnar with palisaded polarized hyperchromatic nuclei and corrugated surface keratin. Proliferating rete ridges in arcading pattern were seen. Features were suggestive of multiple KCOTs. (Fig II)

Based on clinical, radiographic and histopathological examination, and the presence of major and minor diagnostic criteria, a diagnosis of Gorlin Goltz syndrome was made.

The patient was followed up weekly with iodoform packing of the cystic cavities. Healing was uneventful as seen clinically and radiographically. No evidence of recurrence has been noted after follow up for 1 year.

Case 2:

A 13 year old boy, presented with a complaint of swelling over face for the past one and a half months. The swelling was gradually increasing in size, was not associated with pain or pus discharge. Medical history was unremarkable. Examination revealed signs of retarded mental and physical growth. He had increased head circumference, broadened nasal bridge, strabismus, hypertelorism, macrocephaly with frontal bossing, multiple dermal nevi and polydactyly of the left foot

Oral examination revealed a swelling over the right maxillary region that was approximately 3.5 x 3 cms, and was obliterating the right nasolabial fold. It was non-tender, firm-hard with no associated pus discharge or change in color of overlying skin. He had multiple swellings at the right and left body of mandible region. He had a bony swelling in the right maxillary region extending from teeth 11-16 with obliteration of the vestibule and a swelling in the mandibular region with respect to teeth 85, 46 and 74 which was firm, non tender with no associated pus discharge

Radiography showed multiple expansile unilocular cysts in both jaws in relation to unerupted/impacted teeth

Based on clinical examination and radiography a provisional diagnosis of Multiple Odontogenic cysts associated with Gorlin Goltz syndrome was made. (Fig. III)

The patient underwent enucleation of the cystic lesions under general anesthesia. FESS with middle meatus antrostomy was also performed simultaneously.

Histopathology specimens from all sites revealed features suggestive of KCOTs.

The presence of major and minor diagnostic criteria including multiple KCOT's, confirmed Gorlin Goltz syndrome.

The patient is on regular follow up visits and healing has been satisfactory and uneventful.

Table I: Diagnostic Criteria for Gorlin Goltz Syndrome

Major criteria
1. Excessive numbers of basal cell carcinomas out of proportion with prior sun exposure and skin type or < 20 yrs of age
2. KCOT of the jaws prior to 20 yrs of age
3. Palmar or plantar pitting
4. Lamellar calcification of the falx cerebri
5. Medulloblastoma, typically desmoplastic
6. 1st degree relative with Gorlin-Goltz syndrome
Minor criteria
1. Rib anomalies
2. Other specific skeletal malformations and radiologic changes (i.e. vertebral anomalies, kyphoscoliosis, short 4th metacarpals, postaxial polydactyly)
3. Macrocephaly
4. Cleft lip and/or palate
5. Ovarian/cardiac fibroma
6. Lymphomesenteric cysts
7. Ocular abnormalities (i.e. strabismus, hypertelorism, congenital cataracts, glaucoma, coloboma)

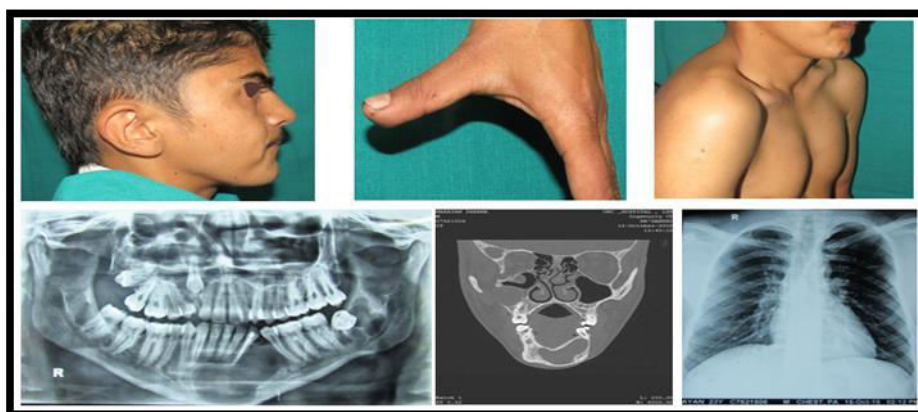


Figure I: Case 1 Clinical and Radiological Findings



Figure II: Case 1 Operative and Histopathological Findings



Figure III: Case 2 Clinical and Radiological Findings

DISCUSSION

Gorlin Goltz [Basal cell nevus syndrome/nevoid basal cell carcinoma syndrome (NBCCS)/basal cell carcinoma nevus syndrome (BCCNS)/Gorlin syndrome] is a syndrome that predisposes to tumour formation, namely basal cell carcinomas, KCOT, as well as skeletal anomalies. Characteristic bony changes consistent with this syndrome were noted in two Egyptian skeletons (3000–2575 BC).^[1] It was first reported by Jarisch and White in 1894 but was described as an autosomal dominant syndrome in 1960 by Dr. Gorlin and Dr. Goltz in a family with basal cell carcinomas, jaw cysts, and bifid ribs.^[2] It is rare with an estimated prevalence varying from 1/57,000 to 1/560,000^[3-7]. Since the first description of the syndrome, the clinical and radiologic findings have been further characterized and diagnostic criteria established.^[5, 7-9] The original criteria as given by Evans et al have been updated and changed at the First International Colloquium on Basal Cell Nevus Syndrome

(BCNS) and have been described in the Consensus statement as given in table I.^[10]

Diagnosis of Gorlin-Goltz syndrome can be made in the presence of: a) 2 major criteria, b) 1 major criteria and molecular confirmation or c) 1 major and 2 minor criteria.^[3] Clinical criteria are quite good in establishing a suspected diagnosis and molecular-genetic confirmation is not warranted in all cases^[10].

The most frequent skin sign of Gorlin-Goltz syndrome are basal cell carcinomas of the face, back and chest. Most basal cell carcinomas appear between puberty and 35 years of age although they have been reported to occur as early as at two years of age.

Palmoplantar pits are another major clinical sign with patients having small 1-2-mm asymmetric palmar/plantar pits. The pits are permanent and increase in number with age. When pits are found in a child, they are a strong diagnostic marker.^[11]

Skeletal abnormalities with skull, rib and vertebral column shape defects are also frequent.

KCOT are the main oral sign and occur in the mandible about three times as often as in the maxilla. Less than 10% of the patients with multiple OKCs have other manifestations of this syndrome. It has been suggested that multiple OKCs alone may be confirmatory of the syndrome ^[11]. In the Gorlin Goltz syndrome, KCOTs occurs at a much younger age and were often the first sign of NBCC in 78% of the cases.

The genetic basis of the syndrome was identified with causative mutations in several genes in the sonic hedgehog signalling pathway, including PTCH1, PTCH2 and SUFU ^[10].

All odontogenic cysts have shown positive immunoreaction for the heparanase protein in various intensities. Their results imply that heparanase expression may be correlated with the neoplastic properties of KCOT, particularly in NBCCS associated cases. ^[12]

Our patients primarily presented with complaints related to the presence of KCOTs. Gorlin Goltz syndrome was diagnosed on the basis of the presence of major and minor criteria. Both patients were treated with enucleation of the lesions with adjunctive therapy of chemical cauterization with carnoy's solution in the mandible. The KCOTs in the maxilla were enucleated and FESS with middle meatus antrostomy was performed in order to allow the maxillary sinus to reepithelialise with specialized epithelium. This was done keeping in mind the aggressive nature and recurrence rate of KCOTs. Since Gorlin Goltz syndrome is a multi system condition, a multi-team approach needs to be applied ^[10]. For our cases, consultations were taken from the Neurosurgery and Radiology departments. The surgery was performed by a joint team of Maxillofacial surgeons and ENT surgeons. The intra oral approach was used by the maxillofacial team for enucleation of the lesions, while an endoscopic approach through FESS and middle meatus antrostomy was performed by the ENT team. Impacted teeth that were located within the sinus and were not visible through the intraoral incision were visualized endoscopically and extracted. Because of this multi-team approach, we could ensure complete removal of the lesions in the maxilla, sinus and mandible.

CONCLUSION

In Gorlin Goltz syndrome, KCOT's are often the first clinical feature to be recognised and could be the only condition for which the patient may seek treatment. Both our cases presented with complaints related to the presence of KCOTs and were diagnosed with Gorlin

Goltz syndrome on the basis of major and minor criteria after detailed examinations.

It is therefore important that OMF surgeons have knowledge of the main features of this syndrome and a high index of suspicion be present when a patient of multiple KCOT presents to our clinics.

REFERENCES

1. Satinoff M Wells C. Multiple Basal Cell Naevus Syndrome in Ancient Egypt. Medical History. 1969;13(03):294-297.
2. Gorlin R Goltz R. Multiple Nevoid Basal-Cell Epithelioma, Jaw Cysts and Bifid Rib. New England Journal of Medicine. 1960;262(18):908-912.
3. Larsen A, Bygum A, Mikkelsen D, Hertz J. Manifestations of Gorlin-Goltz syndrome. Danish Medical Journal. 2014;61(5).
4. Farndon P, Del Mastro R, Kilpatrick M, Evans D. Location of gene for Gorlin syndrome. The Lancet. 1992;339(8793):581-582.
5. Shanley S, Ratcliffe J, Hockey A, Haan E, Oley C, Ravine D et al. Nevoid basal cell carcinoma syndrome: Review of 118 affected individuals. American Journal of Medical Genetics. 1994;50(3):282-290.
6. Lo Muzio L, Nocini P, Savoia A, Consolo U, Procaccini M, Zelante L et al. Nevoid basal cell carcinoma syndrome. Clinical findings in 37 Italian affected individuals. Clinical Genetics. 1999;55(1):34-40.
7. Evans D, Ladusans E, Rimmer S, Burnell L, Thakker N, Farndon P. Complications of the naevoid basal cell carcinoma syndrome: results of a population based study. Journal of Medical Genetics. 1993;30(6):460-464.
8. Kimonis V, Goldstein A, Pastakia B, Yang M, Kase R, DiGiovanna J et al. Clinical manifestations in 105 persons with nevoid basal cell carcinoma syndrome. American Journal of Medical Genetics. 1997;69(3):299-308.
9. Kimonis V, Mehta S, Digiovanna J, Bale S, Pastakia B. Radiological features in 82 patients with nevoid basal cell carcinoma (NBCC or Gorlin) syndrome. Genetics in Medicine. 2004;6(6):495-502.
10. Bree A Shah M. Consensus statement from the first international colloquium on basal cell nevus syndrome (BCNS). American Journal of Medical Genetics Part A. 2011;155(9):2091-2097.
11. Muzio L, Nocini P, Bucci P, Pannone G, Consolo U, Procaccini M. Early diagnosis of nevoid basal cell carcinoma syndrome. The Journal of the American Dental Association. 1999;130(5):669-674.
12. Katase N, Nagatsuka H, Tsujigiwa H, Gunduz M, Tamamura R, Pwint H et al. Analysis of the neoplastic nature and biological potential of sporadic and nevoid basal cell carcinoma syndrome-associated keratocystic odontogenic tumor. Journal of Oral Pathology & Medicine. 2007;36(9):550-554.

Source of support: Nil

Conflict of interest: None declared

This work is licensed under CC BY: *Creative Commons Attribution 3.0 License*.