

Case Report

Gorlin Goltz syndrome: a rare case report

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ABSTRACT

Gorlin-Goltz syndrome is uncommon multisystemic disease with an autosomal dominant trait, with complete penetrance and variable expressivity, though sporadic cases have been described. We report a case of 18 years old male patient having features of Gorlin Goltz syndrome. Gorlin-Goltz syndrome is characterized by multiple basal cell nevi or carcinomas, odontogenic keratocysts, palmar and/or plantar pits, calcification of the falx cerebri, and is associated with internal malignancies. It is important to know the major and minor criteria for the diagnosis and early preventive treatment of this syndrome.

Keywords: Calcification of the falx cerebri, Gorlin-Goltz syndrome, Odontogenic keratocysts

INTRODUCTION

Gorlin-Goltz syndrome is an autosomal dominant disorder characterized by cutaneous basal cell carcinoma, multiple keratocystic odontogenic tumors, and skeletal anomalies.¹ This syndrome is usually diagnosed early by the otolaryngologists because keratocystic odontogenic tumors are usually one of the first and most common manifestations of the syndrome. Early diagnosis is important in reducing the severity of long term manifestations of this syndrome

This syndrome has been termed with several names such as, basal cell nevus syndrome, GGS, nevoid basal cell carcinoma syndrome (NBCCS), multiple basal cell carcinoma (BCC) syndrome, multiple basalioma syndrome, jaw cyst basal cell tumor skeletal anomalies syndrome, jaw cyst bifid rib basal cell nevus syndrome, nevoid basalioma, odontogenic keratocysts skeletal anomalies syndrome and fifth phacomatosis.²

CASE REPORT

A male patient 18 years old presented in dentistry OPD with chief complaint of swelling in bilateral cheeks.

Patient was conscious, co-operative, well oriented to time and place.

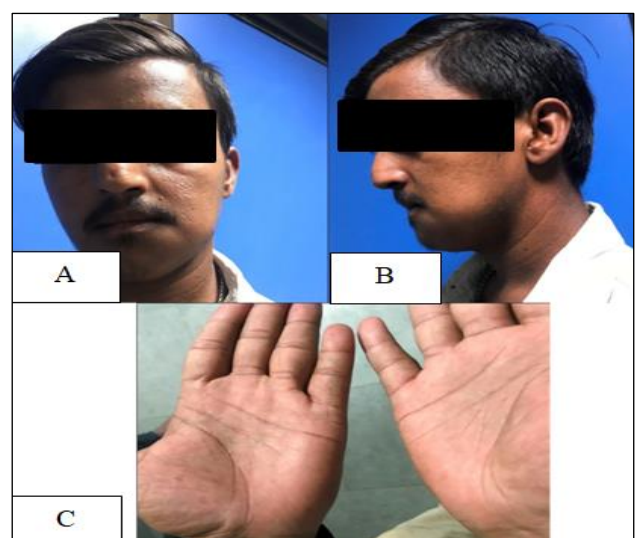


Figure 1: A, B) Frontal bossing, broad nasal bridge, C) Palmar pits.

The duration of the swelling was 6 month and the growth was slow in nature. There was no associated history of pus discharge or trauma. On examination, the swelling was firm and slightly tender. Examination of the face showed mild frontal bossing, prominent supraorbital ridges, widened nasal bridge (Figure 1 a, b). Palmar pits in both hands were evident (Figure 1 c)

On general physical examination, the patient was moderately built and nourished and presented with normal gait, with satisfactory vital signs.

An orthopantomograph (Figure 2) revealed multiple radiolucent lesions on both sides of the maxilla and of the mandible.



Figure 2: Multiple radiolucencies located in maxilla and mandible with displaced teeth.

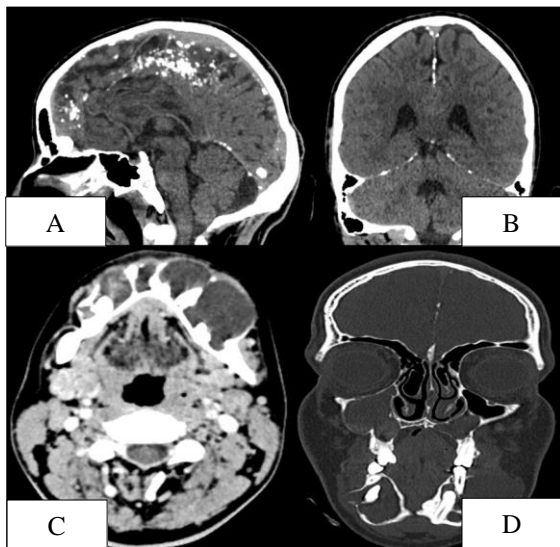


Figure 3: A, B) Plain Computed tomography brain images showing calcification of falx and tentorium cerebelli. C) Contrast enhanced Computed tomography (soft tissue window) image showing expansile lytic lesions in body of mandible. D) Computed tomography (bone window) image showing expansile lytic lesions in body of mandible and in maxilla (the expansile lytic lesion of right maxilla has extended into maxillary sinus through its floor).

For the further evaluation CECT face was advised -

Computed tomography (CT) scan of the brain and face revealed calcification of the falx cerebri (Figure 3 a, b) and multiple cystic lesions (odontogenic keratocysts) in maxilla and mandible (Figure 3 c, d).

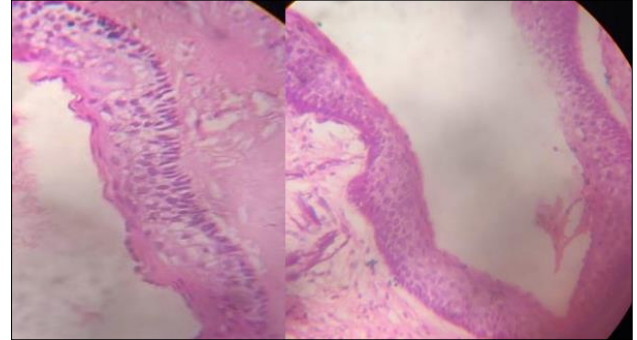


Figure 4: Histopathological picture of the odontogenic keratocyst.



Figure 5: VRT image of bifid right fourth rib and bifid left third and fifth rib.

Multiple cysts in the jaws and findings in extraoral examination were suggestive of Gorlin syndrome, so further evaluation was done, in which presence of bifid ribs was observed (Figure 5).

Based on the diagnostic criteria for nevoid BCC syndrome established by Evans et al, and modified by Kimonos et al, the patient was diagnosed as having Gorlin-Goltz syndrome.^{3,4}

DISCUSSION

The Gorlin-Goltz Syndrome is an autosomal dominant inherited disease. The tumor-suppressor (PTCH) gene, located in the 9q22.3 chromosome is one of the causes of the Gorlin-Goltz syndrome.^{5,6} Mutation of this gene produces dysregulation of several genes. The syndrome includes a wide spectrum of defects involving the skin, eyes, central nervous and endocrine systems, and bones. It is also known as basal cell nevus syndrome.⁷ Diagnosis is based on the most frequent and specific features of the syndrome as given by Evans et al.⁷ Diagnosis can be

established when two major or one major and two minor criteria are present.⁷⁻⁹

The major criteria are

- Multiple (more than 2) basal cell carcinomas or one occurring under the age of 20 years
- Histologically proven OKCs of the jaws
- Palmar or plantar pits (three or more)
- Bilamellar calcification of the falx cerebri
- Bifid, fused or markedly splayed ribs
- First-degree relative with NBCCS.

The minor criteria are

- Macrocephaly (adjusted for height).
- Congenital malformation: Cleft lip or palate, frontal bossing, coarse face, moderate or severe hypertelorism
- Other skeletal abnormalities: Sprengel deformity, marked pectus deformity, marked syndactyly of the digits
- Radiological abnormalities: Bridging of the sella turcica, vertebral anomalies such as hemivertebrae, fusion or elongation of the vertebral bodies, modeling defects of the hands and feet or flame shaped radiolucencies hands or feet
- Ovarian fibroma
- Medulloblastoma.

In our patient, the diagnosis of the GGS was established by the presence of four major criteria (viz., multiple OKC, calcified falx cerebri, bifid ribs, palmar pits) and one minor criterion (viz., frontal bossing, widened nasal bridge). Multiple basal cell carcinomas are considered one of the most common characteristics of GGS, especially in the head and neck region.¹⁰ Another feature of GGS is the occurrence of multiple KCOTs of the jaw.¹¹ The occurrence of GGS-associated KCOTs is approximately a decade earlier than that of KCOTs not associated with the syndrome.¹² KCOTs are the most frequent and consistent features of GGS. KCOTs associated with GGS are more common in the mandible than the maxilla.¹³

CONCLUSION

Early diagnosis of GGS is important to reduce the severity of complications, such as associated malignant tumors. It is important to recognise this syndrome because of its malignant potential. For early diagnosis of NBCCS, clinical and imaging examinations in early ages of life is required. Since the transmission is autosomal dominant with good penetrance there is the need of genetic counseling. The understanding of the syndrome is necessary for early diagnosis and management of cutaneous tumours associated with the syndrome.

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Ethical approval: Not required

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