Gorlin-Goltz Syndrome: Case report

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Abstract
Gorlin Goltz syndrome involves multiple organ systems having high penetrance and variable expressiveness. It is observed that most aggressive forms of odontogenic keratocysts are the first signs of this syndrome which show high recurrence rate due to its varied histopathological features. It is a rare syndrome having an estimated incidence of 1 in 50,000 to 150,000 in the general population with a 3:1 male/female gender predilection. Thus attention is required from oral clinicians to recognize the condition at an early stage so as to render effective treatment. Gorlin-Goltz syndrome has rarely been reported from India. We report here one such patient, who was diagnosed and followed at teaching hospital.

Key words: Gorlin-Goltz syndrome; Multiple odontogenic keratocysts; Palmar plantar keratosis; Hypertelorism.

Introduction
Gorlin-Goltz syndrome is an infrequent multisystem disease that is inherited in a dominant autosomal way, which shows a high level of penetrance and variable expressiveness (1). This syndrome has received several names throughout the times such as "basal cell nevus syndrome", "nevoid basal cell carcinomas syndrome", or the most complex name of "multiple basal epithelioma, jaw cysts and bifid rib syndrome" (2). It is rare autosomal dominant condition and patients with this syndrome often have anomalies of multiple organs, many of which are subtle (Table 1).
A case of Gorlin-Gotz syndrome

Clinicians should be familiar with the features of syndrome and regular follow up after treatment as these patients have increased propensity to develop multiple neoplasm’s, including basal cell carcinoma and medulloblastoma (3) which are life threatening.

Table 1: Clinical manifestations of the syndrome (3, 4)

<table>
<thead>
<tr>
<th>Category</th>
<th>Features</th>
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<tbody>
<tr>
<td>Cutaneous anomalies</td>
<td>Basal cell nevus, other benign dermal cysts and tumors, palmar pitting, palmar and plantar keratosis, and dermal calcinosis.</td>
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<tr>
<td>Dental and osseous anomalies</td>
<td>Multiple odontogenic keratocysts (OKC), mild mandibular prognathism, frontal and temporoparietal bossing, kyphoscoliosis or other vertebral defects, bifurcated ribs, spina bifida, and brachymetacarpalism.</td>
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<tr>
<td>Ophthalmic anomalies</td>
<td>Hypertelorism, wide nasal bridge, dystopia canthorum, congenital blindness, and internal strabismus.</td>
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<tr>
<td>Neurological anomalies</td>
<td>Mental retardation, dural calcification, bridging of sella, agenesis of corpus callosum, congenital hydrocephalus, occurrence of medulloblastoma.</td>
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<tr>
<td>Sexual anomalies</td>
<td>Hypogonadism, ovarian tumor-like fibrosarcoma</td>
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Case report

A 12 year old male patient reported to outpatient department of Vishnu Dental College, Bhimavaram with the complaint of swelling on left side of the face (figure 1) since 2-3 years with no associated pain or discharge except for a feeling of heaviness in the region. History revealed that he was born after a full-term pregnancy and thereafter had an unremarkable medical and dental history. On examination there was hypertelorism with prominent supraorbital ridges, hyperkeratosis of palmar and plantar surfaces of hands and feet (figure 2).

The swelling was single, ovoid in shape and of 3X3 cm in size. On palpation, it was hard in consistency with no fluctuant points and surface changes. Intraoral examination (figure 3) revealed soft fluctuant swelling obliterating the buccal sulcus area in the region of 26, retained 62, 63 and missing 23 & 33. Fine needle aspiration was performed from fluctuant area which retrieved a straw colored fluid as aspirate.

The panoramic view (figure 4) of the patient revealed four areas of well-defined radiolucencies involving the maxillary and mandibular jaws. A unilocular radiolucency in the region of 22 to 27 with impacted 23 and 28, mandibular arch showed presence of unilocular radiolucency involving three areas at 48 and 37, 38 region extending to ramus of
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mandible, another radiolucency seen to extend from 35 to 43 crossing midline and displacing the roots of the teeth in the path with impacted 33.

Figure 3: Bi-cortical expansion obliterating the buccal sulcus area

Figure 4: Panoramic view showing well defined radiolucencies involving multiple sites of mandible

Other radiographs (occlusal views and posterior-anterior & lateral views of skull) disclosed no abnormalities. No other anomalies of the skeletal, cardiovascular, or central nervous system were present. A diagnosis of Gorlin Goltz Syndrome was made following the criteria for diagnosis. The parents of the patient were examined and underwent radiological evaluation; neither of them had any features of the Gorlin-Goltz syndrome. All the cystic lesions of the jaws were enucleated surgically, histopathological examination of tissue showed the features of odontogenic keratocyst (figure 5).

Figure 5: Histopathological report suggestive of odontogenic keratocyst

A regular follow up of every six months with complete rehabilitation was advised and the patient has been followed up for six years (figure 6) with no recurrence or any other features.

Figure 6: Panoramic view taken after period of six years showing no recurrence of lesion

Discussion

Gorlin and Goltz in 1960 established a classical triad (Multiple basocellular epitheliomas, keratocysts in the jaws and bifid ribs) that characterizes the diagnosis of this syndrome (4). This triad was later modified by Rayner et al (5) who established that for giving the diagnosis at least cysts had to appear in combination with calcification of the falx cerebri or palmar and plantar pits or keratosis.
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Diagnosis of the syndrome is made using major and minor criteria which include the following; Major criteria: Multiple basal cell carcinoma or one occurring under the age of 20 years, histologically proven Odontogenic Keratocysts (OKCs) of the jaws, palmar or plantar pits (three or more), bilamellar calcification of the falx cerebri, bifid or fused or markedly splayed ribs and first-degree relative with Nevoid Basal Cell Carcinoma syndrome (NBCCS) (6, 7). Minor criteria: 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 syndactily of the digits. Radiological abnormalities- bridging of the sella turcica, vertebral anomalies (hemivertebrae, fusion or elongation of the vertebral bodies), modeling defects of the hands and feet or flame shaped hands or feet, ovarian fibroma and medulloblastoma (6, 7).

In our patient the diagnosis of the Gorlin-Goltz syndrome was established by the presence of two major criteria (viz., multiple odontogenic keratocysts, palmar plantar keratosis) and minor criteria (viz. hypertelorism).

It has been suggested that multiple OKCs alone may be confirmatory of the syndrome (1). Although benign, the recurrence rate after excision of OKC is high, ranging from 12 to 62.5% and multiple recurrences are not unusual (3). The PTCH 1 gene, the human homolog of the Drosophila segment polarity gene, has been seen to be involved in the development of the NBCCS (8).

Although Multiple OKCs can occur as a part of some rare dermatological syndromes, such as Bazex syndrome (rare acral psoriasiform dermatosis, internal malignancy, carcinoma of upper aerodigestive tract), Torre’s syndrome (skin sebaceous tumours with internal malignancy) (9) but this patient was apparently healthy and had no features suggestive of these syndromes.

The clinical management includes surgical treatment (enucleation and peripheral ostectomy) of the lesions (10). Addressing the risks associated with syndrome, the development of neoplasm’s on sun or radiation exposure, recurrent odontogenic keratocysts etc., warrants utmost importance. Thus a regular follow up, interdisciplinary cooperation for the diagnosis, treatment and rehabilitation is required.

Patients suffering from this syndrome has to undergo check-ups at least once a year, especially the ones having odontogenic keratocysts (2) so our patient was on regular follow up initially for every six months till three years followed by yearly recall for another three years.

**Conclusions**

Our case highlights the importance of the awareness of this rare syndrome especially in young people without any skin lesions. As multiple odontogenic cysts alone may be confirmatory for the diagnosis of this syndrome which is easily recognizable in routine radiographic examination of the head and neck, even as an incidental finding offering the opportunity for its early diagnosis and treatment. A protocol for regular follow-ups which increases the chances for better overall survival rates is required and recommended.

**References**

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