**Case Report**

Gorlin-Goltz syndrome

Munish Kohli, Monica Kohli, Naresh Sharma, Saif Rauf Siddiqui, S. P. S. Tulsi

**ABSTRACT**

Gorlin-Goltz syndrome is an inherited autosomal dominant disorder with complete penetrance and extreme variable expressivity. The authors present a case of an 11-year-old girl with typical features of Gorlin-Goltz syndrome with special respect to medical and dental problems which include multiple bony cage deformities like spina bifida with scoliosis having convexity to the left side, presence of an infantile uterus and multiple odonogenic keratocysts in the maxillofacial region.

**Key words:** Autosomal dominant, multiple organs, odontogenic keratocyst, spina bifida

**INTRODUCTION**

The Gorlin-Goltz syndrome or nevoid basal cell carcinoma (BCC) syndrome or nevus epitheliomatodes multiplex, or nevoid basal-cell epithelioma-jaw cyst-bifid rib syndrome is an uncommon, autosomal dominant disorder affecting multiple organ systems which include skeletal, eye, skin, reproductive, and neural system, although all the features are rarely observed in a single patient.\(^1\) It is principally characterized by cutaneous BCCs (seen in 50–97% of people with the syndrome), multiple keratocysts (present in 75% of people), and skeletal anomalies. The incidence of this disorder is estimated to be 1 in 50,000 to 150,000 in the general population, varying from region to region. Many of the symptoms were first described by Jarish and White in 1894, while in 1960 Robert J Gorlin and Robert W. Goltz defined the condition as a syndrome comprising the principal triad of multiple basal cell nevi, jaw keratocyst, and skeletal anomalies.\(^2\) It appears in all ethnic groups, but most often in Whites; males and females are equally affected.

**CASE REPORT**

An 11-year-old female patient attended the Department of Oral and Maxillofacial surgery, Saraswati Dental College and Hospital, with a chief complaint of delay in the eruption of permanent teeth. A brief medical history revealed that during late infancy (11 months) she was diagnosed to have rickets and treated for the same. Apart from this, her past history was uneventful. Her psychomotor development was normal and her school performance was above average.

At the Department of Oral and Maxillofacial surgery, Saraswati Dental College and Hospital, the patient was completely examined. On inspection, the patient appeared to have sprengel deformity [Figure 1]. AP view of the chest revealed multiple bony cage abnormalities. Significant findings elicited were spina bifida of C5 and C6 vertebrae with scoliosis having convexity to the left side and left and right 4th and 5th ribs partially fused with narrow intercostal space [Figure 2]. Complete blood count was done and appeared to be within normal limits. Panoramic radiograph revealed multiple radiolucent areas in the region suggestive of a cyst [Figure 3]. Aspirates of right ramus region revealed a total protein content of 0.8 g/dl, suggestive of a keratocyst. Enucleation with open dressing (packing the cavity with iodoform gauze) of the right ramus region cyst was done. Enucleation of the left body mandible cyst was done. The cystic lining was sent for histopathologic examination and was confirmed as odontogenic keratocyst.

Hitherto fore, the patient was advised to get an ultrasonography of the abdomen which revealed a
small nodular structure in uterine fossa giving an impression of an infantile uterus. Left ovary could not be visualized.

The patient was also referred to a tertiary care centre – Sanjay Gandhi Post Graduate Institute, Lucknow, for genetic evaluation where karyotyping from blood was done and no detectable abnormality was elicited.

Based on the clinical, radiographic, and histologic findings, and referring to the diagnostic criteria for nevoid BCC syndrome established by Evans et al, and modified by Kimonis et al, in 1997, the patient was diagnosed as having Gorlin-Goltz syndrome.

**DISCUSSION**

Gorlin-Goltz syndrome is an autosomal dominant disorder with high penetrance and variable expressivity. It is caused by the mutations in the patched tumor suppressor gene (PTCH), a human homologue of the *Drosophila* gene mapped to the long arm of chromosome 9q22.3-q31. Gorlin-Goltz syndrome's typical malformative patterns suggest that the main function is to control the growth and development of normal tissues. It is associated with multiple keratocysts in patients in the second decade of their life. In the case presented here, one of the first signs was multiple cystic lesions involving the maxilla and mandible, which have been histopathologically diagnosed as odontogenic keratocysts. The association with odontogenic keratocysts, however, is not clearly understood and appears in more than 90% of the cases. All the other disorders are less frequent. Acral pits that are often overlooked during physical examination have a characteristic dermoscopy with red globules that are mainly distributed in parallel lines inside flesh-colored, irregular-shaped, and slightly depressed lesions. Dermoscopy improves the visualization of these pits.

Despite the name of the syndrome, multiple BCCs occur only in 50% of the cases. They may vary in number from a few to 1000 and range in size from 1 to 30 mm in diameter. BCC most often involves face and non-exposed areas such as the back and chest. Management of superficial BCC without hair follicle involvement can be accomplished by the topical application of 0.1% Retinoin cream, 5% 5-Florouracil cream, 5% Imiquimod cream, cryosurgery, and surgical excision.

The diagnostic criteria for nevoid BCC were established by Evans et al, and modified by Kimonis et al. in 1973. According to them, diagnosis of Gorlin-Goltz syndrome can be established when two major or one major and two minor criteria are present which are described below.

**Major criteria**
1. More than two BCCs or one BCC under the age of 20 years.
2. Histologically proven odontogenic keratocyst of the jaw.
3. Three or more cutaneous palmar or plantar pits.
4. Bifid, fused or markedly splayed ribs.
5. First degree relative with nevoid basal cell carcinomas.
Minor criteria
This consists of any one of the following features.
1. Proven macrocephaly, after adjustment for height.
2. One of the several orofacial congenital malformations: cleft lip or palate, frontal bossing, ‘coarse face’, moderate or sever hypertelorism.
3. Other skeletal abnormalities: sprengel deformity, marked pectus deformity, marked syndactyly of the digits.
4. 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 lucencies of the hands or feet.
5. Ovarian fibroma.
6. Medulloblastoma.

In this article, the authors have presented a case having two major (histologically proven odontogenic keratocyst of the jaw and bifed, fused or markedly splayed ribs) and one minor criteria (sprengel deformity).

Conclusion
Gorlin-Goltz syndrome is an entity that often involves the maxillofacial region. Multiple cyst of jaw may be an indicator of this pathology and may occur as early as 7–8 years of age. It is important to make an early diagnosis and proper management of Gorlin-Goltz syndrome mainly due to its malignant predisposition. The guideline for diagnosis includes a family history, careful oral and skin examination, chest and skull radiographs, panoramic radiographs of the jaws, and pelvic ultrasonography in women. This entity shares its differential diagnosis with cherubism.

Diagnosis and therapy of this syndrome requires a multidisciplinary approach (oral surgeons, dermatologists, surgeons, and neurologists and geneticists). It consists of surgical removal of tumors and adequate treatment of keratocyst. Patients with Gorlin-Goltz syndrome require consistent sun protection. Genetic counseling that considers the genetic risks is advisable for all patients with this syndrome, both familial and sporadic.

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