Gorlin-Goltz Syndrome- A Rare Case Report

Dua Nisha, Kapila Rishabh, Trivedi Ashwarya, Gupta SD, Mahajan Parul

Abstract

Gorlin-Goltz syndrome or Nevoid basal cell carcinoma syndrome is an autosomal dominant disorder with a predisposition to cancer. Features like basal cell carcinoma, odontogenic keratocysts, calcification of falx cerebri, bifid ribs, pits on palms and soles and hypertelorism are evident. Due to importance of oral maxillofacial manifestations of this syndrome, it is fundamental to know its characteristic in order to make diagnosis, early preventive treatment and establish the right genetic advice. A case of this rare disease seen on a 22 year old male patient is presented here, characterized by multiple odontogenic keratocyst, malocclusion of the teeth and other skeletal findings.

Keywords:
Odontogenic keratocyst, Gorlin-Goltz Syndrome, Basal Cell Carcinoma.
**Introduction**

Gorlin-Goltz syndrome also known as Nevoid Basal Cell Carcinoma Syndrome is a rare autosomal-dominant disorder characterized mainly by the presence of multiple basal cell carcinomas, odontogenic keratocysts of the jaw and palmar pits. This syndrome is associated with a wide spectrum of developmental anomalies and neoplasms. Other findings of this syndrome include hypertelorism, palmar and planter pits, calcification of falx cerebri, central nervous system and ocular lesions, cleft lip and palate, mandibular prognathism (class III jaw relationship) and in rare cases ovarian fibromas. A case of Gorlin-Goltz syndrome is presented here in which most of the above mentioned findings are evident.

**Case Report**

A 22 years old male patient visited with the chief complaint of malaligned teeth in upper front teeth region since 8 years. Patient underwent surgery for cleft lip and palate at age of 1 and 1.5 years respectively. Extraoral findings included frontal bossing, hypertelorism and mandibular prognathism (class III jaw relationship) and in rare cases ovarian fibromas. A case of Gorlin-Goltz syndrome is presented here in which most of the above mentioned findings are evident.

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Intraoral examination revealed malocclusion of teeth (Fig. 1). All the vitals were within normal limits. Patient was advised to undergo a panoramic radiograph (Fig. 2) which revealed, on left side a well defined homogeneous multilocular radiolucency with scalloped corticated border, approximately 5 cm in mediolateral dimensions extending from beneath the area of missing 35 and going backwards upto middle 1/3rd of ramus and ends approximately 2cm beneath the sigmoid notch. Superio-inferiorly, it started approximately from the alveolar crest of 37, 38 region and going inferiorly upto lower border of mandible displacing the mandibular canal downward. Also showing impacted 37 and 38 within the radiolucency and resorption was seen i.r.t roots of 37. On right side, another well defined homogeneous unilocular radiolucency with corticated border approximately 2cm in maximum dimension was seen, encircling the coronal portion of 48, present on the anterior border of the right ramus area. The impacted supernumerary teeth were also present in the right and left posterior region of maxilla suggestive of multiple cystic lesions. Differential diagnosis includes Odontogenic keratocyst, Dentigerous cyst, Unicystic Ameloblastoma and odontogenic myxoma.

Due to the presence of multiple cysts like lesions in the jaw, Gorlin-Goltz syndrome was suspected and further investigations were carried out. A radiograph of the skull revealed calcification of falx cerebri (Fig. 3) and a chest radiograph showed bifid ribs (Fig. 4).
Blood investigations were within the normal limits and surgical enucleation of cystic lesion was done from both the right and left side of mandible and histopathological report (Fig.5) revealed the overall features suggestive of odontogenic keratocyst.

So, based on history, clinical examination, radiological examination and histopathological examination, final diagnosis of Gorlin Goltz Syndrome was given and the patient was followed up after every 3 months for a year after the surgery and once a year after that.

**Discussion**

Gorlin-Goltz syndrome was described for the first time in 1894 by Jarisch and White. Later in 1939, Straith described a familial case in which multiple
basocellular carcinoma and cysts appeared. In 1953, Gross presented a case suggestive additional signs such as synostosis of first rib and bilateral bifurcation of sixth rib. On the other hand, Bettley and Ward were the first to relate the presence of palmar and plantar pits with the syndrome. It was not until 1960 when Gorlin and Goltz established a classical triad that characterizes the diagnosis of this syndrome [multiple basocellular epithelioma, keratocyst in the jaws and bifid ribs]. This triad was later modified by Raynee et al who established that for giving the diagnosis atleast cyst had to appear in combination with calcification of falx cerebri or palmar and plantar pits³.

The tumour suppressor gene called 'patched'[PTCH] located in 9q22.3 chromosome has been identified as the cause of Gorlin Goltz Syndrome⁴. The gene is composed of 23 axons and codifies a transmembrane glucoprotein composed of 1447 aminoacids and 12 domains. Gorlin Goltz syndrome is produced due to mutation in gene with the loss of heterozygosity.

The syndrome presents a quite variable estimated prevalence which goes from 1 in 57,000 to 1 in 2,56,000 inhabitants. It is considered that between 30% and 50% of patients who suffer from this syndrome donot know if any of their family members has ever had it.

The presence of two major or one major and two minor criteria is essential for the diagnosis of Gorlin-Goltz syndrome⁵,⁶,⁷ (Table 1).

<table>
<thead>
<tr>
<th>Major Criteria</th>
<th>Minor Criteria</th>
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<tbody>
<tr>
<td>Pigmented basocellular carcinoma</td>
<td>Cardiac/ovarian fibroma</td>
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<tr>
<td>Odontogenic keratocyst</td>
<td>Cleft palate</td>
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<tr>
<td>Palmar-plantar pits</td>
<td>Mandibular prognathism</td>
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<tr>
<td>Ectopic calcification</td>
<td>Meningioma</td>
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<td></td>
<td>High arched eyebrows and palate</td>
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</table>

The major and minor criteria present in our patient were odontogenic keratocyst and bifid ribs with various minor criteria such as cleft palate and lip, mandibular prognathism and falx calcification helping in the diagnosis of Gorlin Goltz Syndrome.

It is of great importance to make an early diagnosis since severity in complications such as skin and malignant tumours can be reduced and so can the destruction and secondary oral maxillofacial deformities of jaw cyst. An early diagnosis is important to give adequate genetic advice.

The management includes removal of tumours either by surgical excision, topical chemotherapies and laser
ablation. With cases of basal cell carcinoma, radiation therapy should be avoided because it causes invasion of basal cell carcinomas years later. Adequate treatment of cysts- removal of large cyst occasionally weakens the remaining bony integrity and places it at risk of pathologic fracture. This can be managed with intermaxillary fixation or placement of reconstruction plate. In case of odontogenic keratocysts, there are different treatment techniques to eliminate and to avoid the high rate of recurrence. The therapeutic techniques for the keratocysts vary from simple enucleation with curettage, to the enucleation with peripheral osteotomy or to osseous resection in block. Genetic counselling that considers the genetic risks is advisable for all patients with this syndrome.

Conclusion

The presence of multiple odontogenic keratocysts should always alert both the radiologist and clinician to look for signs and symptoms that may suggest Gorlin Goltz Syndrome. In case of high index of suspicion, all the organs systems should be screened for additional criteria. Early recognition of this syndrome is of paramount importance to reduce the morbidity and mortality since multiple features have a malignant potential or aggressive behaviour. The latter include multiple recurrent odontogenic keratocysts, which may become large and cause oromaxillofacial deformation and destruction.

References
