Biomedical Research 2010; 21 (4): 355-358

Case Report- Gorlin Goltz Syndrome (GGS) - Report of a rare case.

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Abstract

Gorlin's Goltz syndrome (GGS) is a rare autosomal dominant inherited condition. It involves many organs, but principally affects the skin, skeleton, endocrine and nervous systems. It consists of the classic triad of basal cell carcinomas, multiple jaw cysts and skeletal deformities along with other defects. The authors report an unusual case of a fifteen year old male patient who reported with the complaint of swelling and mild pain in the lower right side of the jaw. Investigations revealed multiple cystic lesions in both upper and lower jaws which were found to be Odotogenic Keratocysts (OKC), presence of bifid rib and multiple nevi. The clinico-pathological diagnosis was made as Gorlin- Goltz syndrome (GGS) since the present case exhibited three major criterias.

Key words: Nevoid basal cell carcinoma syndrome, Gorlin Goltz syndrome, Bifid ribs, Multiple odontogenic keratocyst cysts, Multiple Nevi, Palmar pits, PTCH gene.

Accepted March 21 2010

Introduction

Gorlin-Goltz syndrome (GGS) is a rare autosomal dominant disorder that involves multiple organ systems, including the skin, skeleton and jaws. Odontogenic keratocysts. bifid ribs and nevoid basal cell carcinomas are the hallmark of the disease.[1-4] Jarisch White in 1894 first reported regarding the association of nevoid basal cell carcinoma with skeletal anomalies as cited by Fitz Patrick PJ.[2] It was in 1960, Gorlin and Goltz established a classical triad that characterises the diagnosis of this syndrome which included multiple basal cell carcinomas, keratocysts in the jaws and bifid ribs and hence the name.[3] We present a rare case of Gorlin Goltz syndrome.

Case Report

A male patient aged 15 years reported to clinics of Jaipur Dental College, Jaipur, with a complaint of swelling on the right lower jaw since one year. The swelling slowly progressed to the present state which the patient noticed only when visible facial asymmetry occurred.

On extra oral examination there was facial asymmetry because of a solitary diffuse swelling on the right side of the face involving middle and lower third region of the face (Figure 1). Intra oral examination revealed that the buccal and lingual vestibule in relation to 44 to 47 was obliterated with diffuse swelling (Figure 2). A thorough oral examination also revealed missing 32, 33, 34 and 35 and retained 74 and 75. Orthopantomograph (OPG) was advised to arrive at a final diagnosis. The OPG revealed multiple multilocular radiolucencies in relation to 47 and 48 and also in relation to impacted 32, 33, 34 and 35. Unilocular radiolucency was also observed in relation to erupting 18, 28 and 38 (Figure 4).

Then out of suspicion, face and other parts of the body were thoroughly examined which revealed multiple nevi on the face, neck and back region (Figure 1). It was decided to go for a chest radiograph as he exhibited pigeon shaped chest (Figure 4). The chest radiograph confirmed the presence of bifid ribs (Figure 5). His hands and feet were searched for any palmar and plantar pits which were not found. CT scan was advised for proper treatment planning. The findings of the CT scan revealed multiple cystic lesions in both the jaws.

Surgical treatment with enucleation of all cystic lesions was done under general anaesthesia with peripheral ostectomy and chemical cauterisation with Carnoy's solution (Figure 6). All the five samples of the cystic lesions along with the involved teeth were sent for detailed histo-

pathological investigations to Department of Oral Pathology (Figure 7). Based on the histological features it was diagnosed as Odotogenic Keratocyst (Figure 8). Similarly other two specimens were also diagnosed as infected Odotogenic Keratocyst with daughter cyst, one with Odontogenic Keratocyst and Dentigerous cyst. Correlating with the clinical, radiographic and histo-pathological features, it was diagnosed as Gorlin Goltz syndrome.

Figure 1: *Photograph showing facial asymmetry and multiple nevi on face and neck*

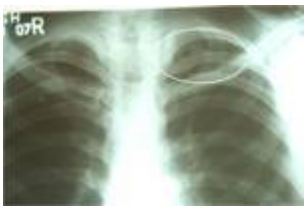

Figure 5: *Chest X-ray showing Bifid rib (circled)*

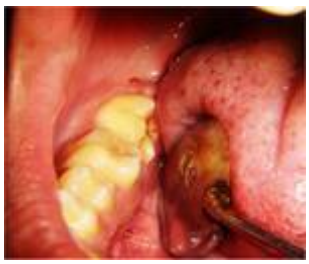

Figure 2: *Intra-oral photograph showing lingual swelling in relation to mandibular posterior region*

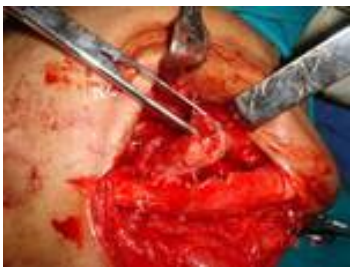

Figure 6: *Intra-operative photograph showing cyst enucleation*

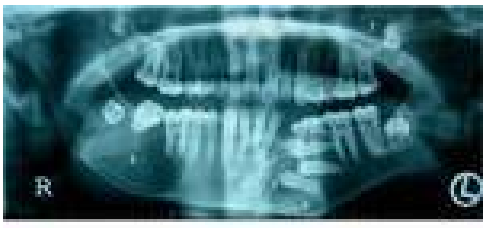

Figure 3: *Orthopantogram showing multiple cystic lesions*

Figure 7: *Gross appearance of all the cystic lesions*

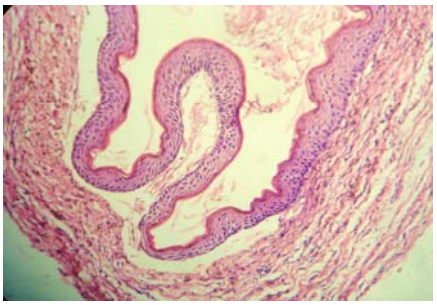

Figure 8a: *Photomicrograph of OKC showing cystic lumen lined by conjugated parakeratinized epithelial lining and capsule (H & E staining 4x)*

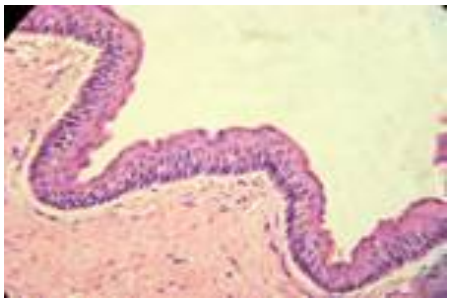

Figure 8b: *Photomicrograph of OKC showing corrugated parakeratinized epithelial lining and seperation from the capsule (H & E staining 20x)*

Discussion

Gorlin-Goltz syndrome is an autosomal dominant inherited disorder with high penetrance and variable phenotype expressiveness which can manifest itself spontaneously.[4,5] It is characterized by the development of multiple odontogenic keratocysts, frequently beginning in the second decade of life, and or basal cell carcinomas (BCCs) usually from the third decade onwards. Approxi mately 60% of individuals have a recognizable appearance with macrocephaly, bossing of the forehead, coarse facial features, and facial milia.

Most individuals have skeletal anomalies (e.g., bifid ribs, wedge-shaped vertebrae). Ectopic calcification, particularly in the falx ceribri, is present in more than 90% of affected individuals, cardiac and ovarian fibromas occur in approximately 2% and 20% of individuals respectively. Approximately 5% of children with GGS develop medulloblastoma [6].

The prevalence of Gorlin syndrome varies from one country to another. 1: 55,600 in England,[7] in Australia it was 1:1,64,000,[8] 1 :13,939,393 in Korea,[9] 1:2,56,000 in Italy.[10] The frequency of the NBCCS has been variously estimated, but there is more or less general agreement that the prevalence is about 1:60,000.[11] Gorlin syndrome is caused by germ-line mutations of the *PTCH* gene.[4] The human homolog of the Drosophila segment polarity gene *Patched* (PTCH) located in the 9q22.3-9q31 chromosome is responsible for the development of GGS. [5]

Gorlin Goltz syndrome is diagnosed in individuals with two major criteria and one minor criterion or one major and three minor criteria (Evans et al).[1] Kimonis et al [12] modified Evans criteria to include more minor criterias.

Since the syndrome is a hereditary condition, thus referral to a geneticist for counselling is mandatory. It is recommended to involve families in regular screening. It may be possible to prove whether someone in a family has inherited the condition or not by DNA tests, either by tracking the chromosome 9 containing the faulty gene, or by direct analysis of the mutation causing the disease in a particular family.

In our case, patient had three major criterias such as multiple odontogenic keratocysts, bifid rib and multiple Nevi and hence it was diagnosed as Gorlin Goltz syndrome.

Early diagnosis of the syndrome is essential to treat it effectively and thereby minimising the complications. Interdisciplinary cooperation of oral and maxillofacial surgeons, oral pathologists, paediatricians, dermatologists, and specialists in genetics is mandatory in the diagnosis, treatment and follow-up of patients with Gorlin Goltz syndrome.

Diagnostic Criteria for Gorlin Syndrome [12].

Major criteria
1. More than 2 basal cell carcinomas or one under the age of 20 years.
2. Odontogenic keratocysts of the jaw proven by histology.
3. Three or more palmar or plantar pits.
4. Bilamellar calcification of the falx cerebri.
5. Bifid, fused or markedly splayed ribs.
6. First degree relative with NBCC syndrome
Minor criteria
Any one of the following features:
1. Macrocephaly determined after adjustment for height.
2. Congenital malformations: cleft lip or palate, frontal bossing, ‘‘coarse face,’’ moderate or severe 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.

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