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GORLIN GOLTZ SYNDROME AFFECTING THE MAXILLA AND MANDIBLE: A CASE REPORT

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ABSTRACT

Gorlin-Goltz syndrome, a rare inherited syndrome occuring in an autosomal dominant manner, presents with basal cell carcinomas, odontogenic keratocysts (OKCs), and skeletal and dermal anomalies. This case report describes occurrence of this syndrome in a 12-year-old female patient, causing distortion of lower jaw. Prompt identification and intervention are vital to mitigate the potential for grave complications, such as malignancies of the skin and brain, and significant disfigurement or destruction of the oral and maxillofacial region due to cysts in the jaw.

INTRODUCTION

Gorlin-Goltz syndrome (GGS) is also referred to as nevoid basal cell carcinoma syndrome (NBCCS) and is inherited as an autosomal dominant disorder. It occurs most commonly in Caucasian population, although it affects all racial groups and both genders are affected equally. The clinical features are seen in the first to third decades of life, affecting multiple organ systems. [1] It occurs due to mutations in the patched-1 (PTCH-1) gene, a tumor suppressor gene found on the long arm of the 9th chromosome. It was first described by Jarisch and White in the year 1894. It was named after Robert W. Goltz and Robert J. Gorlin who in 1960 described the various features of the syndrome, including multiple basal cell carcinomas, odontogenic keratocysts of the jaw and musculoskeletal malformations. [2]

CASE REPORT

A 12 year old female patient reported to the Department of oral and maxillofacial surgery, King George's Medical University, Lucknow with the chief complaint of swelling in the left side of lower jaw since 1 month. The patient gave no history pus or blood discharge from the lesion.

On extraoral examination, gross facial asymmetry was present due to swelling present over lower one third of face on the left side, measuring approximately 1.5 x 2.5 cm in diameter, extending superoinferiorly from ala tragus line till lower border of mandible and anteroposteriorly from the corner of mouth till the left angle of mandible. On palpation the swelling was firm, non tender, with a smooth surface.

Overlying skin was afebrile. Submental and submandibular lymphadenitis was absent.

On intraoral examination, a swelling was noted obliterating the buccal vestibule, extending from the first premolar till the first molar.

High palatal vault, mandibular prognathism, and prominent palatine ridges were also noted.

Other features noted extraorally were mild hypertelorism and frontal bossing (Refer to Figure 1a and 1b)

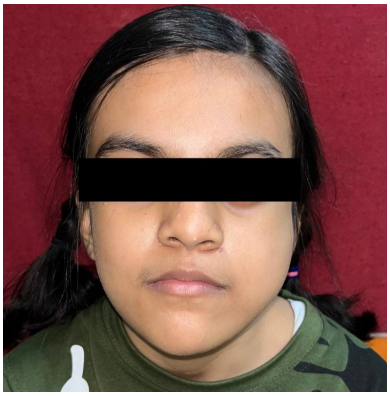

Figure 1a. Pre-operative clinical photographs

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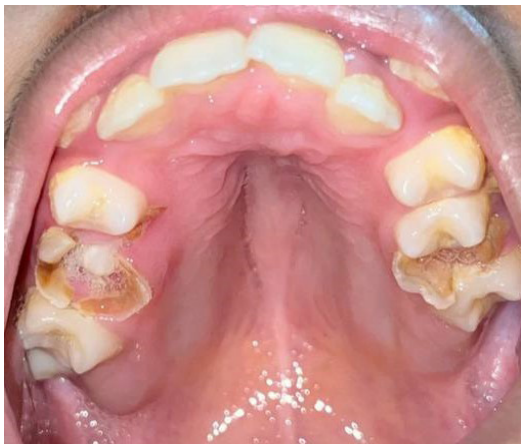


Figure 1b. Pre-operative clinical photographs

Orthopantomography revealed multiple radiolucent lesions bilaterally involving both the maxilla and mandible, causing hollowing out of body and ramus regions bilaterally. Impacted teeth were seen in relation to each of the cysts. Due to the presence of multiple cysts of the jaws and other intraoral findings, Gorlin Goltz syndrome was suspected and further investigations were carried out. (Figure 2)

3D Reconstruction of Computed Tomography of the face showed expansion of buccal cortices of mandible bilaterally leading to gross facial asymmetry. (Figure 3)

Computed Tomography of the brain revealed calcification of the falx cerebri and PA Chest revealed presence of bifid ribs. (Refer to Figure 4a and 4b)

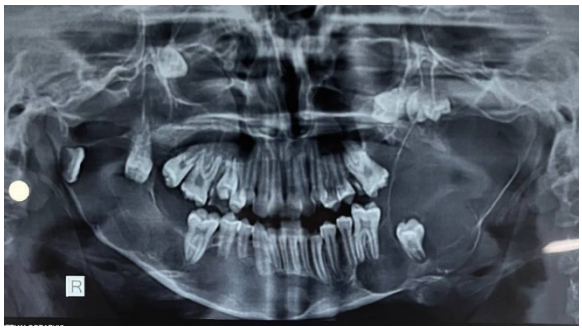


Figure 2. Orthopantomography of the face

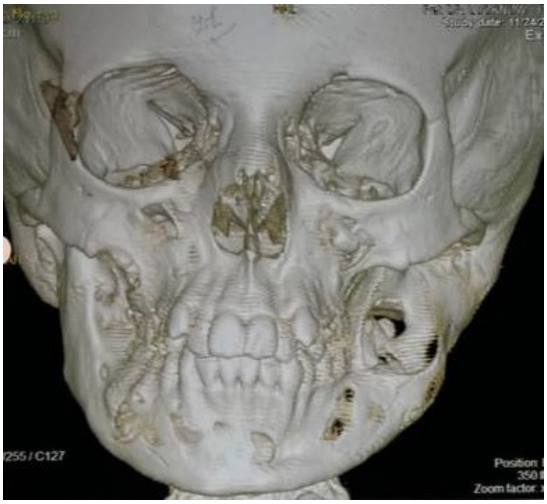
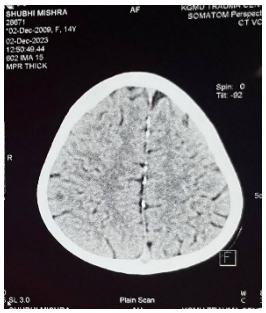


Figure 3. 3D Reconstruction of CT Face



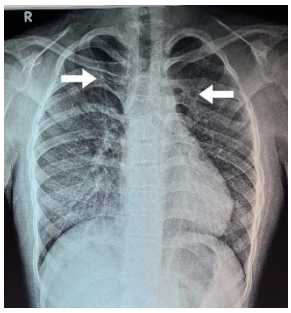


Figure 4a and **4b**. Calcification of falx cerebri and bifid ribs

Routine blood investigations were normal.

Aspiration of the cystic contents was done and sent for biochemical analysis which revealed low albumin, high protein and high sugar content.

MANAGEMENT

Cystic enucleation was done under general anesthesia via intra oral approach.

The enucleated cystic lining from each lesion was sent for histopathological examination, revealing a 6-8 cell layer thickened epithelial lining having hyperchromatic nuclei. At various foci artefactual clefting between epithelium and underlying fibrocollagenous tissue was seen. Underlying zones showed dispersed keratin debris. Few foci showed bony trabeculae. Thus, confirming the diagnosis of Odontogenic Keratocysts.

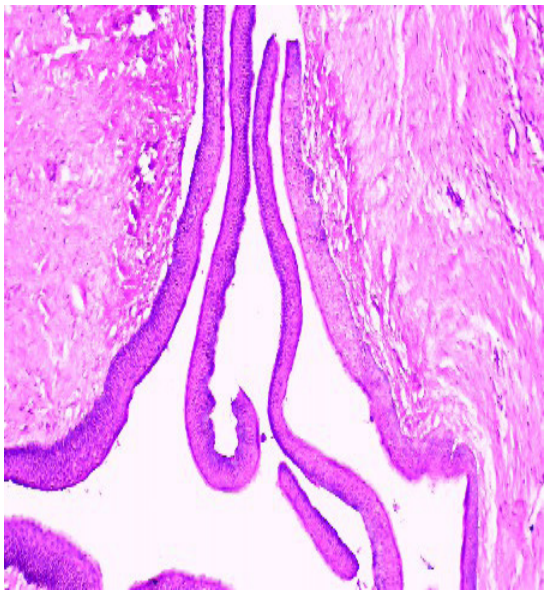


Figure 5. Histopathological examination of the lesion

OUTCOME AND FOLLOW UP

Regular follow up was done and the patient experienced an uneventful postoperative recovery, with no complications noted during follow-up. She continues to do well without signs of recurrence or infection

Figure 5. Post operative clinical photograph

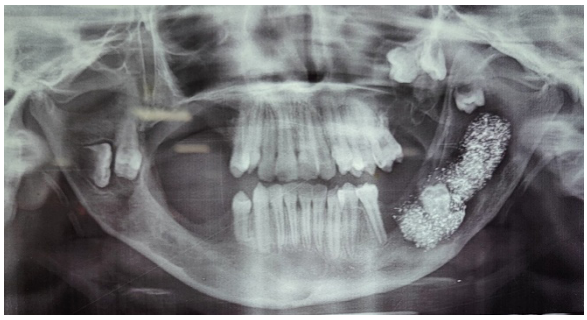


Figure 6. Post operative OPG of the patient

DISCUSSION

Gorlin Goltz syndrome Is an uncommon genetically acquired disorder with a variety of clinical features. involves the skin, nervous system, eyes, endocrine system, and skeleton. It was first reported by Jarisch and White in 1894 in a patient with multiple BCCs, scoliosis and learning disability. [3] Multiple terms are used to refer to this syndrome which are- basal cell nevus syndrome, multiple basal cell carcinoma syndrome, Gorlin syndrome, multiple nevoid basal cell epithelioma-jaw cysts, or bifid rib syndrome.

Gorlin and Goltz reported the presence of the classical triad consisting of multiple basal cell epitheliomas, keratocysts in the jaws, and bifid ribs which confirmed the diagnosis of the syndrome, later changed by Rayner et al to add simultaneous presence of flax cerebri calcification or palmoplantar pits. [4]

Further modifications were introduced by Evans et al in 1993 and Kimonis et al in 1997 which led to the establishment of following diagnostic major and minor criteria:

Major criteria

- Multiple Basal Cell Carcinomas or one lesion occurring under the age of 20 years
- Histologically proven OKCs of the jaws
- Palmoplantar pits (three or more)
- Bilamellar calcification of the falx cerebri
- Bifid, fused or markedly splayed ribs
- First degree relative with NBCCS.

Minor criteria

- Macrocephaly
- Congenital malformations (Cleft lip / palate, coarse facies, frontal bossing, moderate to severe hypertelorism)
- Other skeletal abnormalities: Sprengel deformity, pectus deformity, syndactyly of the digits
- Radiological abnormalities: Bridging of the Sella turcica, vertebral anomalies such as hemivertebrae, modelling defects of the hands and feet or flame shaped hands or feet, fusion or elongation of the vertebral bodies
- Ovarian fibroma
- Medulloblastoma

Patient should fulfil 2 major criteria or 1 major and 2 minor criteria for diagnosis.

In this case, three major criterias were met- Falx cerebri calcification, bifid ribs and histologically proven odontogenic keratocysts of jaw.

The incidence of GGS is assessed to be 1 in 50,000 to 150,000 in general population, depending on the region. [5] It has both a sporadic and a familial incidence and although spotted in very young children, they are commonly reported between the ages of 17 years and 35 years. [6]

Woolgar et al. and Dominguez et al. [7] found considerable differences between syndromic keratocysts and non-syndromic keratocysts. Syndrome keratocysts were found to have a distinctly increased quantity of satellite cysts, solid islands of epithelial proliferation, odontogenic rests within the capsule, and mitotic figures in the epithelial lining of the primary cavity.

Mutations of the PTCH1 gene, which is part of the hedgehog – signaling pathway, is the molecular cause of the syndrome. Mutations in this gene results in loss of control of several genes known to play a role in organogenesis, carcinogenesis and odontogenesis thus resulting in the development of GGS. [8]

The management of OKCs can be either conservative or aggressive. In young patients , conservative treatment methods should be always considered first because an aggressive treatment can cause unfavourable results on the development of jaw, tooth development, and the eruption process. Most preferable method is marsupialisation followed by enucleation.

Aggressive treatment or en bloc resection of the lesion is considered only when there is recurrence or there is aggressive clinical behaviour. [9]

The guidelines for follow-up of NBCCS as given by de Amezaga et al [10] should be followed:

- Neurological examination – twice yearly
- Cerebral MRI –yearly for 1-7 years of age
- Skin examination – yearly
- Cardiologic examination – according to sign and symptoms
- Genetic counselling of families as it is an autosomal dominant disorder

CONCLUSION

In GGS, an early diagnosis is of major importance in order

for prompt detection and management of long term and potentially fatal complications such as skin and brain malignancies. Prompt diagnosis also helps in avoidance of maxillofacial deformities due to the jaw cysts

It is vital to differentiate a sporadic cyst from a syndromic cyst as the recurrence rate varies considerably, therefore the treatment plan might differ.

Patients identified with GGS should undergo dermatological examination every 3–6 months and a once in a year OPG should be advised to look for new cysts or recurrence

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