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Case Report

Gorlin-goltz syndrome: A case report

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ABSTRACT

Introduction: Gorlin- Goltz syndrome is an inherited autosomal dominant disorder. This shows a high level of penetrance and variable expressiveness. Early diagnosis of this syndrome is important for counseling of patients to prevent harmful exposure to Ultra-Violet and ionizing radiations that increase the risk of developing Basal Cell Carcinoma.
Presentation of Case: A 18 years old male patient came with chief complaint of swelling in lower front region of jaw. Clinical examination done. Orthopantomograph revealed multiple multi-locular, well-defined radiolucency with sclerotic border located in maxilla and mandible. Relevant investigations were done including chest X-ray, Computed Tomography scan and Histopathological examination revealed presence of Gorlin- goltz syndrome.
Discussion: The Gorlin-Goltz syndrome is manifested by multiple defects involving the skin, nervous system, eyes, endocrine system, and bones. The presence of two major and one minor or one major and three minor criteria are necessary to establish diagnosis. The present reported case of Gorlin-Goltz syndrome fulfilled 3 major and 1 minor criteria.
Conclusion: In the treatment of recurrent Odontogenic Keratocyst, associated with Gorlin- goltz syndrome, the overlying surface epithelium should be excised along with the cystic lining to prevent recurrence. This paper focus on the importance of oral and maxillofacial health professionals in the early diagnosis of Gorlin-Goltz syndrome and in a multidisciplinary approach to provide a better prognosis to the patient.

1. Introduction

The Gorlin-Goltz Syndrome (GGS) is inherited autosomal multisystemic disease. It is also known as the nevoid basal cell carcinoma syndrome (NBCCS). In year 1894, the Jarisch and White first describe patients of this syndrome, describing the presence of multiple basal cell carcinomas. In 1960 Gorlin and Goltz established the classical triad which characterizes the diagnosis of this syndrome (multiple basal cell epitheliomas, kerato-cysts in the jaws, and bifid ribs) which were then modified by Rayner et al., should be present odontogenic kerato-cysts in combination with calcification of the falx cerebri or palmar and plantar pits.

It was observed that the wide range of other neurological, ophthalmic, endocrine, and genital manifestations are in association with this triad. Male is to female ratio is equal. Currently, genetic studies point out the markers such as PTCH1, PTCH2, and SUFU which are accountable of syndrome. Out of them, mutation in Patched1 (PTCH1) gene has been identified as the main cause for GGS. [1–5]

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2. Case Report

An 19 years old, male patient came to Oral & Maxillofacial Surgery department, Rural Dental College, Loni, had given chief complaint of swelling in lower front region of jaw. The duration of the swelling was 3 months & was of slow growing nature. No significant history of tooth ache in same region. No history of pus discharge or bleeding from the lesion.

On examination, well defined extra oral swelling seen on lower one-third of face on the right side. Extending supero-inferiorally from 2.5 cm below ala-tragus line to lower border of mandible and antero-posteriorly from corner of mouth to 4 cm ahead of angle region of mandible. Measuring approximately 3 x 3 cm. Overlying mucosa was normal. On the palpation the swelling was firm in consistency, non-tender, afebrile.

On examination, frontal bossing, broad nasal bridge, hypertelorism noticed. Orthopantomograph showed multiple well - defined multilocular radiolucencies, with presence of sclerotic border in maxilla and mandible. Further, the multiple cysts in the jaws, and the extra-oral examination point out towards Gorlin syndrome and so other confirmatory & relevant investigations were done including Radiographs, CT scan and Histopathological examination.

Fig. 1: Presence of frontal bossing, broad nasal bridge and hypertelorism

Flecks and keratin whorls seen in specimen during histopathological examination. There is evidence of cystic lining suggestive of- Odontogenic Keratocyst. Further, enucleation of the cysts was carried out surgically in the Oral & Maxillofacial surgery department, Rural Dental College, Loni. Other skin lesions like basal cell nevus or keratosis were absent.

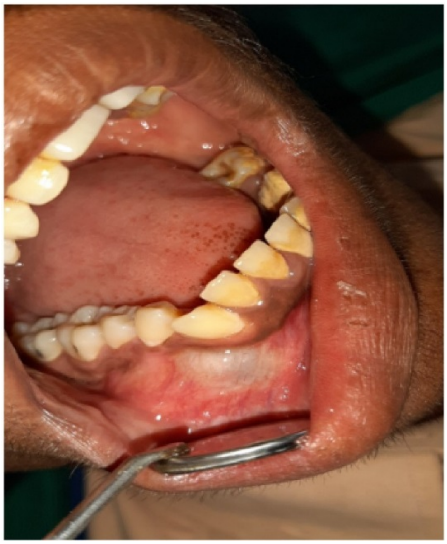

Fig. 2: Shows intra oral swelling in buccal vestibule in canine region

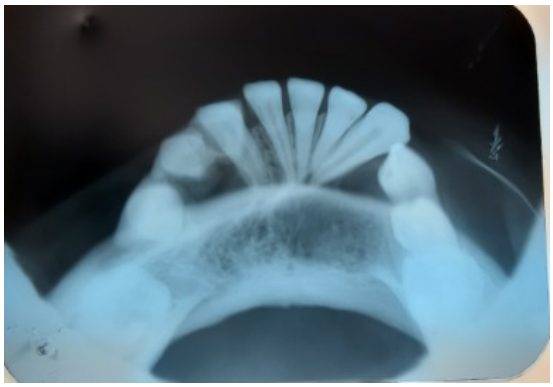

Fig. 3: Occlusal radiograph showing well defined radiolucency and pathological migration of teeth.

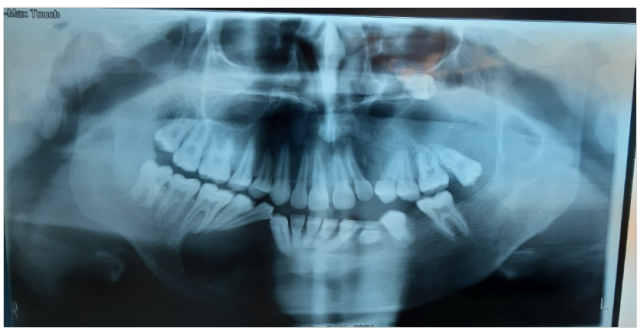

Fig. 4: Panoramic radiograph shows multiple well-defined multilocular radiolucencies with presence of sclerotic border into the maxilla and mandible.

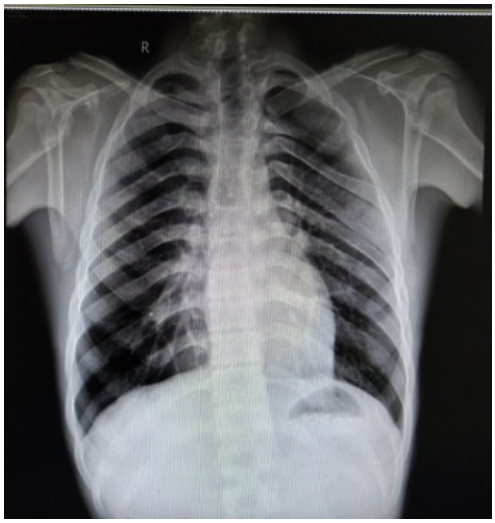

Fig. 5: Chest radiograph showing a bifid right fourth rib.

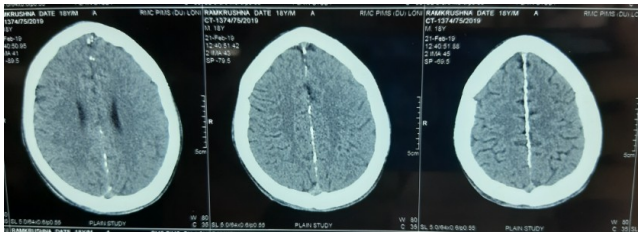

Fig. 6: Calcification of falx cerebri in computed tomography sca

Based on the clinico-radiographic, & histological findings, with reference of the diagnostic criteria, patient was diagnosed with the GGS.

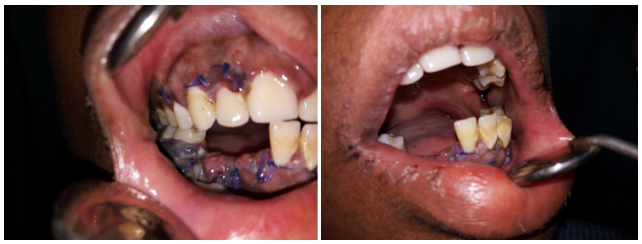

Fig. 7: The incisional biopsy was advised from the lesion. Presence of abundant keratin

3. Discussion

In Gorlin-Goltz syndrome, which is an autosomal dominant inherited syndrome, which involves the skin, nervous system, eyes, endocrine system, and skeleton. The condition is also called as the basal cell nevus syndrome, multiple basal cell carcinoma syndrome, Gorlin syndrome, multiple nevoid basal cell epithelioma-jaw cysts, or hereditary cutaneo-mandibular polyonocosis or bifid rib syndrome. To set up a diagnosis of the Gorlin–Goltz syndrome, the criteria selected were given by Evans et al. (1993), which was later modified by Kimonis et al. (2004). Diagnosis of GGS is confirmed by fulfilling two major and one minor or one major and three minor criteria.[6–11]

3.1. Major criteria for GGS

1. Basal Cell Carcinoma (multiple or one occurring under the age of 20 years)
2. Histopathologically proven KCOTs of the jaws
3. Three or more palmar or plantar pits
4. Bilamellar calcifications of the falx cerebri
5. Bifid, fused, or markedly splayed ribs
6. First-degree relative with GGS.

3.2. Minor criteria for GGS

1. Macrocephaly
2. Cleft lip or cleft palate and other congenital malformation
3. Frontal bossing, coarse face, and moderate or severe hypertelorism
4. Other skeletal abnormalities: Sprengel deformity, marked pectus deformity, and marked syndactyly of the digits
5. Radiological abnormalities: Bulging of sella turcica, vertebral anomalies such as hemi vertebrae, fusion, or elongation of vertebral bodies, defects of the hands and feet, or flame-shaped hands or feet.
6. Ovarian fibroma
7. Medulloblastoma

The Woolgar et al. and Dominiguez et al. reported remarkable distinguishing points between single and syndrome keratocysts. The Syndrome keratocysts had noticeable increase in number of satellite cysts, solid islands of epithelial proliferation, odontogenic rests within the capsule, and mitotic figures in the epithelial lining of the main cavity.

4. Conclusion

To conclude, GGS is a dominant autosomal genetic disorder, which is needs to be evaluated by Craniofacial health experts. Early diagnosis of the condition is important for counselling of patients to avoid harmful exposure from UV and ionizing radiations which increase the chances to develope Basal Cell Carcinoma. For the early diagnosis of GGS, experts should suggest clinico-radiographic examinations in early stages of life.

5. Source of Funding

None.

6. Conflict of Interest

None.

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