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CASE REPORT

Gorlin-Goltz Syndrome with Multiple OKC in a 10-Year-Old Child: A Case Report

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ABSTRACT

Gorlin-Goltz syndrome is rare multi-system disease, which is, characterized by neoplasms and other developmental abnormalities.[1] It is a hereditary condition inherited as an autosomal dominant trait and caused by abnormalities in the PTCH1 (Patched1) gene which is traced to the long arm of chromosome 9q22.3-q31. It is characterized by the triad of multiple baso-cellular epitheliomas, odontogenic keratocysts (OKC) in the jaws and skeletal anomalies. Early diagnosis and treatment are important for long-term prognosis of the syndrome by reducing the severity of cutaneous carcinomas and deformities due to jaw cyst. Current case discusses a 10-year-old child suffering from Gorlin-Goltz syndrome, this case report emphasizes on early diagnosis and prompt treatment of such case.

Keywords: OKC; Gorlin-Goltz syndrome; Multi nodular radiolucency

1 INTRODUCTION

Odontogenic keratocyst (OKC) is an odontogenic cyst of developmental nature that is considered to have originated from the dental lamina, and often associated with an impacted or unerupted tooth/tooth bud. In 1956, Phillipsen was the first to note it. OKC is easily identified due to its rapid growth, aggressive nature, and high tendency to perforate the neighbouring tissues including bone. Its recurrence tendency, at times is associated with basal cell nevus syndrome or Gorlin Goltz Syndrome.[1,2]

Gorlin-Goltz syndrome (GGS) as identified by Gorlin and Goltz in 1960, is a rare genetic disorder that presented with a classical triad of Multiple basal cell nevi, Skeletal anomalies and Odontogenic keratocysts (OKC). It has tremendous penetrance with a myriad of radiologic or dermatologic observations as well as numerous variants of neoplasms.[3,4]

GGS is caused by the mutation of tumor suppressor gene PTCH-;1 present in the long arm of the chromosome 9q22.3-q31 and hence autosomal dominant inheritance is characteristic of GGS[1,3,5]

2 CASE REPORT

A 10-year-old Female patient reported to Department of Pediatric and Preventive Dentistry, Vinayaka Missions Sankarachariyar Dental College, Salem, Tamil Nadu, India, with chief complaint of slowly growing swelling in the lower right front tooth region for the past 4 days. Patient also gave a history of pain while brushing. Pain that is localized, intermittent and dull in nature, aggravates on touching and it relives spontaneously. Patient gives medical history of eye

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surgery before 5 yrs for strabismus correction with no post-operative complications. Right Submandibular lymph nodes are palpable, soft in consistency and non-tender. Other regional lymph nodes are not palpable.

On Extra oral Examination of Swelling, a diffuse swelling is seen in the lower 1/3rd of the face measuring 6cm*4cm extending anteriorly crosses the midline from Para symphysis up to mesial side of 85. Posteriorly extends 5cm away from the angle of the mandible. Superiorly extends from the lower border of the lower lip upto the base of the mandible inferiorly. Surface is smooth, colour is normal to adjacent structures. No bleeding or sinus opening present. On Palpation swelling was tender on palpation with no discharge**.** Evident Frontal Bossing and disproportionate Rule of 5 for facial proportions and hypertelorism (Figure 1).

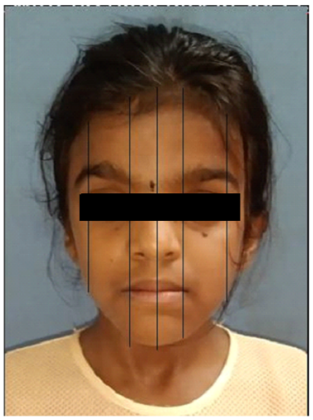

Fig. 1: Frontal Profile Image showing frontal Bossing and disproportionate Rule of 5 for facial proportions

On Intra oral Examination, a diffuse swelling was seen on the lower labial vestibule measuring about 4*3 cms extending from mesial side of 41 to distal side of 84. Blanching noted in relation to 41. Dilation of veins seen. Surface is smooth. No pus discharge is seen (Figure 2). On Palpation all inspectory findings were confirmed by palpation. The swelling is Compressible, non-fluctuant and tender on palpation. Consistency is soft to firm. Soft in the region of 41, 83. No bleeding or pus discharge seen.

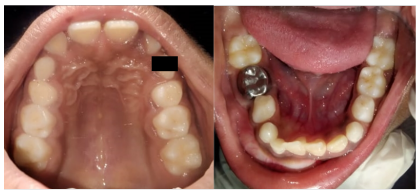

Fig. 2: Pre-op Intra-oral Images. Evident Buccal cortical expansion and vestibular obliteration noted irt 31 to 46, crossing the midline

OPG and CBCT (Figure 3) revealed multi nodular radiolucency seen in the apical region of 41 extending to the resorbed roots of 82, 83 and 84. And radiolucency extends the lower border of the mandible. In the posterior left body of mandible, presence of radiolucency is seen in relation to impacted 37. Radiolucency extends from distal side of 36 to the crown of 37 and from the upper border of the mandible to 2 cms away from the lower border of the body of the mandible.

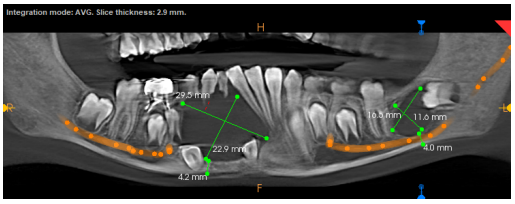

Fig. 3: CBCT i mage showing evident multiple Multi nodular radiolucency

Radio-opaque teeth like structures seen in the lower border of the mandible suggestive of Impacted 42 and 43. Roots of 41 are distally tilted and 31 are mesially tilted.

Fine Needle Aspiration shows cheese coloured fluid with small course granules. Also, cytology report is suggestive of inflammatory cyst with Protein content 2.0g/dL* (Figure 4).

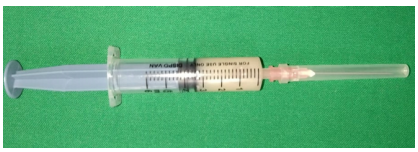

Fig. 4: Fine Needle Aspiration shows cheese coloured fluid with small course granules

Biopsy along with marsupialization of both lesion sites done (Figure 5) under GA. Involved teeth were also extracted to prevent recurrence. Residual surgical site treated with Carnoy's Solution dipped gauze followed by copious irrigation with Normal Saline. Wound Closure done using 3-0 Braided Black Silk Sutures, and haemostasis was verified, post-surgical splint (Figure 6) was place before successful extubation of the patient.

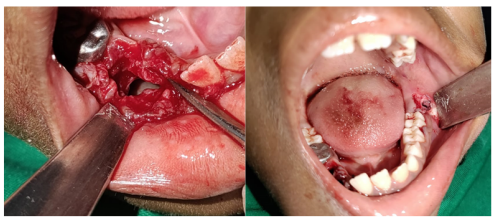

Fig. 5: Marsupilization with evacuation of cystic contents

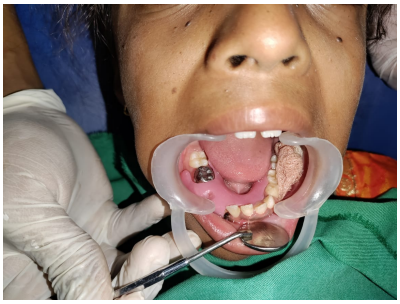

Fig. 6: Pre-fabricated Post-surgical Splint placement done

Histopathological slides (Figure 7) showed Para keratinized stratified squamous epithelium thrown into finger like projections, Para keratin plugging is seen in between the projections. The underlying connective tissue is scanty, thin, and projecting into the epithelial projections into the hyperplastic epithelium. Patient is on regular follow.

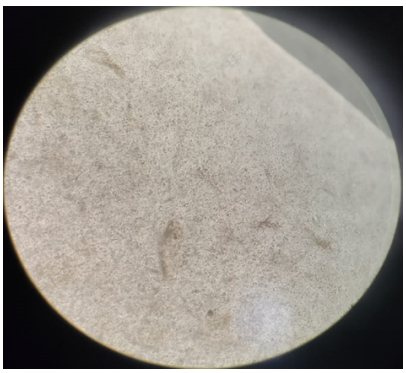

Fig. 7: Histopathological section of the biopsy

3 DISCUSSION

In the present case report, it is possible to confirm the presentation of the signs and symptoms of this syndrome within the second decade, noting that one of the main presentations noted is keratocysts, more commonly within the second to fifth decades.

The diagnostic requirement for the Gorlin-Goltz syndrome has been divided into major and minor requirements, the first includes the presence of more than two basal cell carcinomas or a history of diagnosis of the Gorlin-Goltz syndrome in a first-degree relative; three or more palmoplantar pits; odontogenic keratocysts of the jaw (histologically confirmed); bifid, fused, or markedly splayed ribs; and one basal cell carcinoma below the age of 20 years. The minor requirements are macrocephaly, cleft lip palate, falx cerebri calcification, congenital anomalies, coarse face, frontal bossing, skeletal anomalies (rib anomalies, kyphosis/scoliosis, neurologic or central nervous system anomalies (medulloblastoma, meningioma, agenesis of the corpus callosum, congenital hydrocephalus, and intellectual disability), shortened fourth metacarpal, hemivertebra and combined vertebral corpi, polydactyly/syndactyly, radiolucencies/pseudocysts in the hands or feet, sloping shoulders, and immobile thumbs), impacted or ectopic teeth, hypertelorism, bilateral coronoid hyperplasia and oligodontia sprengel deformity, bridging of the sella turcica, pectus deformity, medulloblastoma, and ovarian fibroma. The specific confirmation of the Gorlin-Goltz syndrome is attained by the availability of two major requirements or two minor and one major requirements. [1–3,6,7]

The features mainly Multiple OKC of jaw, presence of frontal bossing, hypertelorism, sloping shoulders, presence of impacted or ectopic teeth helped us in coming to the conclusion that the patient is suffering from Gorlin Goltz syndrome.

The identification of this syndrome is a complicated, specifically in kids, as the bulk of the major criteria are not evident until the second or third decade. [7]

4 CONCLUSION

Odontogenic keratocysts is a benign pathology of odontogenic origin causing about 10% of most odontogenic cysts and specified by an aggressive nature. Although radiographs don't necessarily provide a conclusive diagnosis, the information on typical and atypical radiological picture of OKCs is necessary for its identification and treatment [8].

Cases like this showcase the necessity for awareness about this rare syndrome in children not having any skin pathology. Prior identification of syndrome and periodic follow-up is necessary cause of possible severity of clinical presentation. Also, a multidisciplinary unit is necessary, especially a dentist, geneticists, neurologist, and dermatologist, to improve outcome and survival rates. In this case, Gorlin-Goltz syndrome got identified and operated for one of its major manifestations, i.e., OKCs, whereas the further manifestations didn't necessitate urgent procedures at this point. [9]

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- **Conflicts of interest/Competing interests:**

No conflict of interest.

REFERENCES

1) Kamil AH, Tarakji B. Odontogenic Keratocyst in Children: A Review. *Open Dentistry Journal*. 2016;10:117–123. Available from: http://dx.doi.org/10.2174/1874210601610010117.
2) Kimonis VE, Goldstein AM, Pastakia B, Yang ML, Kase R, Digiovanna JJ, et al. Clinical manifestations in 105 persons with nevoid basal cell carcinoma syndrome. *American Journal of Medical Genetics*. 1997;69(3):299–308. Available from: https://pubmed.ncbi.nlm.nih.gov/9096761/.

3) Rafiq S, Manzoor F, Dar MA, Aslam R. Imaging of Gorlin - Goltz syndrome: Series of 2 cases. *Journal of Oral and Maxillofacial Pathology*. 2021;25(2):373. Available from: httpa://doi.org/10.4103/0973-029X.325261.
4) Manfredi M, Vescovi P, Bonanini M, Porter S. Nevoid basal cell carcinoma syndrome: A review of the literature. *International Journal of Oral and Maxillofacial Surgery*. 2004;33(2):117–121. Available from: https://doi.org/10.1054/ijom.2003.0435.
5) Chaudhary S, Sinha A, Barua P, Mallikarjuna R. Keratocystic odontogenic tumour (KCOT) misdiagnosed as a dentigerous cyst. *BMJ Case Reports*. 2013;2013:1–5. Available from: https://doi.org/10.1136/bcr-2013-008741.
6) Kimonis VE, Mehta SG, Digiovanna JJ, Bale SJ, Pastakia B. Radiological features in 82 patients with nevoid basal cell carcinoma (NBCC or Gorlin) syndrome. *Genetics in Medicine*. 2004;6(6):495–502. Available from: https://doi.org/10.1097/01.gim.0000145045.17711.1c.
7) Lima FBDJB, Viana APC, Lima LHF, Ribeiro BC, Dutra CEA, Stabile GAV, et al. A Rare Case of Gorlin-Goltz Syndrome in Children. *Case Reports in Dentistry*. 2019;2019:1–5. Available from: https://doi.org/10.1155/2019/1608783.
8) Borghesi A, Nardi C, Giannitto C, Tironi A, Maroldi R, Bartolomeo FD, et al. Odontogenic keratocyst: imaging features of a benign lesion with an aggressive behaviour. *Insights Imaging*. 2018;9(5):883–897. Available from: https://doi.org/10.1007/s13244-018-0644-z.
9) Tandon S, Chauhan Y, Sharma M, Jain M. Gorlin-Goltz Syndrome: A Rare Case Report of a 11-Year-Old Child. *International Journal of Clinical Pediatric Dentistry*. 2016;9(3):264–268. Available from: https://doi.org/10.5005/jp-journals-10005-1374.