International Journal Dental and Medical Sciences Research
Volume 3, Issue 6, Nov-Dec 2021 pp361-364www.ijdmsrjournal.com ISSN: 2582-6018

GorlinGoltz Syndrome with History of Tuberculosis

ManjunathVijapur[1], MDS, AyshaKaleem Pasha[2*], MDS,Vasanth Kattimani[3], MDS, MujahidAhmed [4], MDS,Nidhi Jenson Ukken,[5] MDS,Tejesh Yelamali[6] ,MDS.

[1]Associate Professor, Department of Oral and Maxillofacial Surgery, KIMS, Hubballi, Karnataka, India

[2,5] Post graduate, Department of Oral and Maxillofacial Surgery, KVGDCH, Sullia, DK, Karnataka, India

[3] Specialist, Department of Oral and Maxillofacial Surgery, KIMS, Hubballi, Karnataka, India

[4]Assistant Professor, Department of Conservative &Endodontics, SSDC, Tumkur, Karnataka, India

[6] Consultant, Oral & maxillofacial Surgery, Hubli , Karnataka, India

***Corresponding author**

Submitted: 01-12-2021 Revised: 12-12-2021 Accepted: 15-12-2021

ABSTRACT:

Introduction:Gorlin-Goltz syndrome is a hereditary autosomal dominant disease with full penetrance and a wide range of expressivity. The maxillofacial odontogenickeratocyst (OKC) is a frequent developing odontogenic cyst.

Case Report: A patient reported to our department with complaints of swelling on the mandible on the right side. He was clinically diagnosed with multiple odontogenickeratocyst. This article emphasises the significance of diagnostic criteria, surgical management, his history of tuberculosis and patient prognosis in GorlinsGoltz syndrome cases. Odontogenickeratocyst is notorious for high recurrence; therefore, a proper surgricalenucleation and follow up will determine the outcome of the surgery.

Discussion: Gorlin-Goltz syndrome is a multisystem disease characterized by basal cell nevi, jaw keratocysts, and skeletal abnormalities. The maxillofacial odontogenickeratocyst (OKC) is a frequent developing odontogenic cyst. This trio is known to be linked with a variety of additional neurological, ophthalmic, endocrine, and genital symptoms. The condition is diagnosed using both major and minor criteria. Surgical management of these patients is a necessity in improving their prognosis of the diseases.

Conclusion: The necessity of oral and maxillofacial health experts in the early diagnosis and surgical care of Gorlin-Goltz syndrome is emphasized in this study.

Key Words: Multiple OdontogenicKeratocyst, GorlinGoltz syndrome, Tuberculosis, BIPP pack, Oral & maxillofacial surgery

I. INTRODUCTION

Gorlin-Goltz syndrome is an autosomal dominant disease with varying expressivity and a high degree of penetrance [1, 2, 3]. It is caused by chromosomal sub-band 9q22.3 micro-deletions affecting the patched-1 (PTCH-1) gene, a

tumorsuppressor gene involved in the Sonic Hedgehog (SHH) pathway for cell differentiation. [4]

Jarisch and White published the first reports of individuals with this condition in 1894, emphasizing the presence of numerous basocellular carcinomas. Nonetheless, it wasn't until 1960 that Gorlin and Goltz defined the traditional triad that characterises the diagnosis of this condition (multiple basocellularepitheliomas, keratocysts in the jaws, and bifid ribs). This trio is known to be linked with a variety of additional neurological, ophthalmic, endocrine, and genital symptoms. The general population's incidence of this condition is believed to be 1 in 50,000 to 150,000; however, the perceived prevalence may vary by location. Males and females are both impacted. [5]

It is distinguished by basal cell carcinomas, odontogenickeratocysts, palmar and/or plantar pits, and ectopic calcifications of the falxcerebri. To establish a diagnosis, two major and one minor criterion must be present, or one major and three minor criteria must be present. Its clinical characteristics appear in the first, second, or third decade of life, affecting numerous organ systems such as the skeletal, ocular, skin, reproductive, and brain systems, however, all of the signs are seldom found in a single patient.

The peculiarity in our case was the history of tuberculosis the patient presented with. This led us to a differential diagnosis of tuberculosis infected cyst, the other differential diagnosis being dentigerous cyst, residual cyst, and radicular cyst.

The reported incidence of oral involvement in tuberculosis patients ranges from 0.05% to 3.65%, when determined by clinical examination, and 20% when determined by postmortem examination. [6]

II. CASE REPORT

A 21- years-old male patient with a complaint of swelling in right side of his upper & lower jaw since one year was referred to the Oral and maxillofacial unit . (Figure 1)

Figure 1: Frontal view

He gave a history of swelling with respect to his anterior maxillary region on right side & a swelling with respect to posterior mandibular region on right side which was insidious in origin, and gradually progressed to present size. He complained of intermittent pain with respect to the swellings. He visited a dental clinic about a year ago for the same.

He gave a past medical history of tuberculosis and was on treatment for 6 months the same about 4 years ago.

In March 2021, he presented to Department of general medicine with complains of cough and expectoration. An ECG was done which revealed sinus tachycardia. He was also suspected to have deranged thyroid levels. However, his thyroid levels were well within normal range except for a minimal high free T4 levels. His sinus tachycardia settled with no significance. He was managed conservatively.

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Clinical evaluation revealed swelling with respect to posterior mandibular region on right side.

He had missing right permanent canine and a retained deciduous canine.(Figure 2) He also had missing a 3rd molar on lower right quadrant.

On general examination plantar pits were seen on left foot. (Figure 3)

Figure 2: Retained deciduous canine

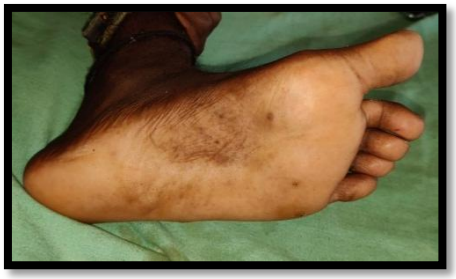

Figure 3: Plantar pits

On admission in the department in August 2021, his systemic examinations revealed no abnormalities and other blood investigations were within normal limits. Sputum test for acid fast bacilli test was negative.

The radiograph of chest was unremarkable and no cutaneous abnormality was revealed.

CT of head and neck revealed cavity measuring about 4 x 2.7cm at posterior right lower border of mandible and a cavity measuring about at 1.2x1.1cm right unerupted maxillary canine region. A smaller cyst was measured about 9x8mm at the left 3rd molar of mandible.

Regarding the CT examination and presence of unerupted teeth and their location, the initial differential diagnosis was dentigerous cyst and the second was KCOT. Other odontogeniccysts and tumors such as adenmatoidodontogenic tumor and tuberculosis infected cyst were considered as other differential diagnoses.

Patient was planned for cyst enucleation under GA. Under all aseptic conditions retained deciduous 53 was extracted. Maxillary vestibular incision was given to reveal impacted 13, which was extracted followed by complete cyst enucleation along with lining. Further 2-3mm of bone was removed to prevent reoccurrence.

An extraoral incision was given with respect to lower border of mandible on right side anterior to the swelling. Blunt dissection was carried out, mental nerve and vessels were identified and preserved. A bur hole was drilled, followed by cyst enucleation revealing a casseous material. (Figure 4)

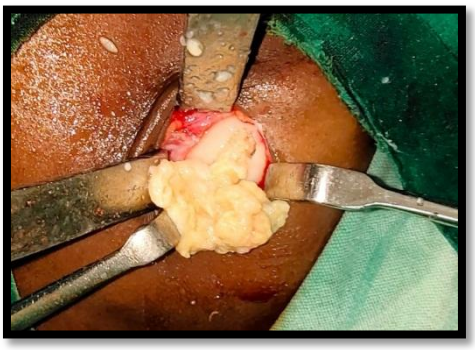

Figure 4: casseous material from lower border of mandible

Tissue samples were sent for histopathology examination. The surgical specimens were sheet-like with casseous cystic appearance. After processing, the tissue samples were sectioned and stained with hematoxylin and eosin (H&E).

(Figure 5)

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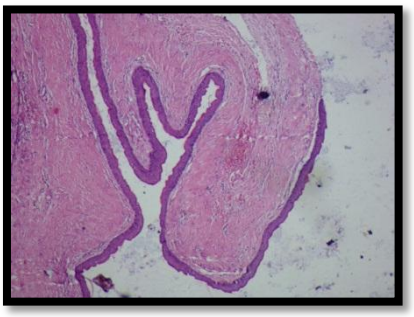

Figure 5: Parakeratinisedodeontogenickeratocyst

The histopathological examination confirmed it to be parakeratinised OKC.

III. DISCUSSION

The Gorlin-Goltz syndrome is an autosomal dominant hereditary disease characterized by numerous skin, neurological system, ocular, endocrine system, and bone abnormalities. Basal cell nevus syndrome, multiple basal cell carcinoma syndrome, Gorlin syndrome, or hereditary cutaneomandibularpolyonocosis, multiple nevoid basal cell epithelioma-jaw cysts, or bifid rib syndrome are all names for this condition. [6]

Jarisch and White reported it originally in 1894, and Gorlin and Goltz expanded on it subsequently.[7]The clinical symptoms of the condition are classified into five groups.

- Cutaneous abnormalities include basal cell nevus, benign dermal cysts and tumours, palmar pitting, palmar and plantar keratosis, and dermal calcinosis.
- Ophthalmic abnormalities include hypertelorism, a broad nasal bridge, dystopia canthorum, congenital blindness, and internal strabismus.
- Neurological abnormalities include mental retardation, dural calcification, sellar bridging, corpus callosum agenesis, congenital hydrocephalus, and the presence of medulloblastoma.
- Sexual abnormalities include hypogonadism and ovarian tumor-like fibrosarcoma.

Clinically, this disease is distinguished by a variety of indications and symptoms. The most common and specific characteristics of the condition, as described by Evans et al. in 1993, are used to make the diagnosis. [8] When two main or one major and two minor criteria are present, Gorlin-Goltz syndrome might be diagnosed.

The following are the major criteria:

1. Multiple basal cell carcinomas or one occurring before the age of 20.
2. Jaw OKCs that have been confirmed histologically.
3. Pits on the palmar or plantar surfaces (three or more).
4. The falxcerebri'sbilamellar calcification.
5. Ribs that are bifid, fused, or spread widely.
6. Nevoid Basal Cell Carcinoma Syndrome in a First-Degree Relative

The following are the minor criteria:

1. Macrocephaly is a condition in which a person's head is (adjusted for height).
2. Cleft lip or palate, frontal bossing, coarse face, moderate or severe hypertelorism are examples of congenital malformations.
3. Other skeletal anomalies include sprengel deformity, pectus deformity, and digit syndactyly.
4. Radiological abnormalities include bridging of the sellaturcica, hemivertebrae, fusion or elongation of the vertebral bodies, and modeling deformities of the hands and feet, such as flame-shaped hands or feet.
5.Ovarian fibroma is a kind of ovarian cancer.
6. Medulloblastoma. [9]

In this case the patient presented with 2 major criteria – multiple odontogenic cyst &

plantar pits and one minor criteria – hypertelorism.

Odontogenickeratocyst.

An odontogenic cyst lined with keratinized stratified squamous epithelium was initially described as an 'odontogenickeratocyst' in 1956. The term 'odontogenickeratocyst,' which is synonymous with 'primordial cyst,' was coined by the World Health Organization (WHO) in 1992 to describe benign cysts of odontogenic origin with a particular histological appearance.However, in 2005, WHO categorized this disease as a benign keratocysticodontogenictumor due to the high risk of recurrence, aggressive clinical history, mutations in the tumor suppressor gene (PTCH1), the presence of satellite cysts, and the connection with the Gorlin–Goltz syndrome (KCOT). The World Health Organization published a revised categorization of head and neck cancers in 2017. Because there was insufficient evidence to classify the aforementioned disease as a neoplastic lesion, KCOT was reclassified as an odontogenickeratocyst (OKC).

The term 'keratocysticodontogenictumour', on the other hand, is still in use. The authors of the 2017 categorization provide no particular recommendations for OKC therapy. Nonetheless, it has been discovered that conservative surgical treatment is not always associated therewith. [10]

OKC were reclassified from benign odontogenictumours to odontogenic developing cysts as a consequence of fresh data about their morphology and biological activity. A mutation in the PTCH1 gene, for example, has been proven to be OKC-specific, as it also occurs in follicular cysts.PTCH1 changes are seen in 30–85 percent of OKCs, depending on whether the lesion is spontaneous or linked with the Gorlin–Goltzsyndrome. It has also been discovered that OKCs retreat after decompression or marsupialization, and that their lining spontaneously transitions into the normal oral epithelium. All of these characteristics prohibit this clinical entity from being classified as a neoplastic lesion and warrant altering the nomenclature of the lesion to OKC. [11]

A unique finding in this case was the history of tuberculosis. Patient gives history of tuberculosis four to five years ago. He underwent treatment for the same. The history of tuberculosis diverted the attention towards a differential diagnosis of tuberculosis infected odontogenickeratocyst.

A remarkable case of primary TB of the mouth is documented, with chronic discharge of pus from the lower wisdom tooth area. An incisional biopsy revealed characteristics of an infected dentigerous cyst, but histological analysis of the excised lesion indicated a keratinizing cyst with secondary infection. Curettage of the region was performed due to non-healing of the bone defect, and the submitted sample microscopically revealed granuloma with typical Langhans' giant cells, indicating the possibility of underlying systemic TB. It was reported at the Subharti Dental College's Department of Oral Pathology and Microbiology in Meerut, Uttar Pradesh. [6]

Systemic and local factors both contribute to the formation of oral TB lesions. Immunosuppression and pathogen pathogenicity are two examples of systemic factors. Poor dental hygiene, local trauma, chronic inflammations, tooth eruption, surgical lesions, periodontal disease, caries, pulp exposure, cysts, and tooth abscesses are examples of local causes. [12]

The treatment of the OKC is still debatable. Treatments are typically divided into two categories: cautious and aggressive. Simple enucleation, with or without curettage, utilising spoon curettes of marsupialization, is a common conservative therapy. Peripheral ostectomy, chemical curettage with Carnoy's solution, and resection are common aggressive treatments. Some surgeons feel that if the lesion is removed whole, the cyst can be adequately treated by enucleation. However, owing to the thin, friable epithelial lining, restricted surgical access, the surgeon's competence and experience, cortical penetration, and the need to preserve surrounding critical structures, full removal of the OKC can be challenging. The therapeutic aim should be to eliminate the possibility of recurrence while also limiting surgical morbidity. There is no agreement on the appropriateness of this lesion's therapy. [13]

The surgical management in this case involved intraoral approach for enucleation for maxillary cyst. In the mandible extraoral approach was used due to size of the lesion and ease of approach to the lesion. This aided in complete removal of lesion with bloodless field, enhancing visibility & accessibility to the surgeon.

The OKC is now being treated with cautious and vigorous enucleation with continuous monitoring. As a method of minimising recurrence, John and James described enucleation in combination with a chemical cauterising agent and excision of underlying mucosa.[14]

IV. CONCLUSION:

GorlinGoltz syndrome is a rare hereditary autosomal dominant disease that should be treatable with early detection and surgical intervention. One of the key characteristics of Gorlin-Goltz is the likelihood of recurrence of odentogenickeratocyst. Proper surgical management and long-term patient monitoring are required, for desired prognosis. A through history shall lead to underlying systemic diseases.

CONFLICT OF INTEREST

None.

AUTHOR CONTRIBUTION

All the authors participated in diagnosing the case & were a part of the surgery and wrote the manuscript. All authors read and approved manuscript.

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