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ORIGINAL RESEARCH

PREVALENCE OF GORLIN-GOLTZ SYNDROME AMONG PATIENTS WITH ODONTOGENIC KERATOCYSTS- A RETROSPECTIVE STUDY.

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ABSTRACT

Background: Odontogenic keratocyst (OKC) is a distinct developmental odontogenic cyst of the jaws known for its characteristic behavior. It has a significant association with Gorlin-Goltz syndrome, which is an autosomal dominant genetic disorder. This study aims to assess the association between OKC and Gorlin-Goltz syndrome and to estimate the prevalence of the syndrome in patients with OKC.

Methods: A retrospective study was conducted among patients who reported to the Department of Oral Pathology at Tamil Nadu Government Dental College and Hospital, Chennai over a 15-year period (2018-2022). Patients with a histopathological diagnosis of OKC were included. Clinical, radiographic findings along with histopathological diagnosis were recorded. Based on the available data, the patients were diagnosed as having Gorlin-Goltz syndrome or not based on the criteria available for identifying the syndrome.

Results: The study population consisted of 120 patients, with a male-to-female ratio of 1.26:1. The mean age at diagnosis of OKC was 40 years. Among the patients with odontogenic keratocysts, 6.6% exhibited clinical features consistent with Goltz-Gorlin syndrome.

Conclusion: The findings indicate that the diagnostic criteria for Gorlin-Goltz syndrome may be expressed differently across various demographic groups.

***Keywords*:** Odontogenic Keratocyst, Goltz-Gorlin Syndrome, Major and Minor Criteria, Multiple jaw Cysts.

INTRODUCTION

Odontogenic keratocyst (OKC) represents a relatively common cystic lesion, comprising approximately 10-20% of all odontogenic cysts[1,2]. It is notably distinguished from other cystic entities by its aggressive clinical course, distinctive histopathological characteristics and a marked tendency for recurrence[1-5.] Odontogenic keratocysts can present either as solitary lesion or as multiple lesions when associated with Gorlin-Goltz syndrome[5-7]. Gorlin-Goltz syndrome is a rare autosomal dominant genetic disorder that present with systemic manifestations affecting multiple organs and tissues throughout the body[8,9]. Notably, the development of odontogenic keratocysts is among its earliest clinical manifestations. These cysts frequently emerge during early childhood or adolescence and in some cases, may represent the initial clinical indicator of the syndrome[10-12]. The early onset, multiplicity of lesions and high recurrence rate of OKCs are critical diagnostic features, not only as a dental concern but also as a potential indicator of broader systemic involvement.

Gorlin-Goltz syndrome exhibits variable degrees of systemic involvement, the extent of which is influenced by the specific genetic mutations occurring during embryonic development[10,13,14]. With no specific diagnostic methods for the identification of the syndrome, the diagnosis of Gorlin-Goltz syndrome is guided by the identification of major and minor criteria[6,15,16]. Literature has shown population-specific variability in the clinical expression of the diagnostic criteria for Gorlin-Goltz syndrome[17-22]. The estimated prevalence of this syndrome has found to be different among various populations, ranging from 1:55000 to 1:256000, with an even lower reported incidence in the Indian population [19,22,23,24]. Most of the existing literature from India comprises of case series, which reveal notable regional differences in systemic involvement among various groups[19,22,25-27].

In this context, the present retrospective study was aimed to evaluate the spectrum of systemic manifestations associated with Gorlin-Goltz syndrome among individuals with a histopathological diagnosis of odontogenic keratocyst, which could contribute to an understanding of its prevalence within this geographic region.

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MATERIALS AND METHODS

A retrospective study was conducted in the Department of Oral Pathology at Tamil Nadu Government Dental College and Hospital, Chennai, Tamil Nadu. Patient records spanning a 15-year period (2008-2022) were retrieved. The permission to conduct this study was cleared from the Institutional Ethical and Review Board. This study included all patients with a histopathological diagnosis of odontogenic keratocyst, irrespective of the patient's age, gender or the number of lesions present. Individual characteristics such as the age, gender, family history, relevant medical and dental history were systematically documented. Particular attention was given to the age at the initial presentation of the cyst, given the recurrence nature of the lesion.

For the odontogenic keratocysts patients, clinical records were systematically reviewed, with attention given to the anatomical localization of the cysts, their association with impacted teeth along with the radiographic changes. The department clinical documentation was thoroughly analyzed to evaluate the systemic involvement in patients diagnosed with odontogenic keratocysts and relevant data were extracted. Cases exhibiting systemic manifestations were identified based on predefined criteria, for the identification of Gorlin-Goltz syndrome and were subsequently recorded. They were categorized as having the syndrome or not based on the identification criteria suggested by Evans and Kimonis et al. In instances where the available information was insufficient, the patients were categorized as not having Gorlin-Goltz syndrome and the details were recorded separately. The collected data were subjected to descriptive statistical analysis to assess the prevalence and patterns of systemic involvement associated with OKC in this part of the country.

RESULTS

The data collected from the Department of Oral Pathology at Tamil Nadu Government Dental College and Hospital, Chennai, Tamil Nadu for a period of 15 years (2008-2022) showed 120 patients with a histopathological diagnosis of odontogenic keratocyst. Of the 120 patients, 67 (56%) were males and 53 (44%) were females with a ratio of 1.2:1. OKC was reported in patients ranging from 11 months to 72 years of age, with both extremes observed in female individuals. The mean age of patients reported with OKC was 40 yrs of age, with the highest peak in the third decade followed by the second decade with a slight male predominance (Fig. 1).

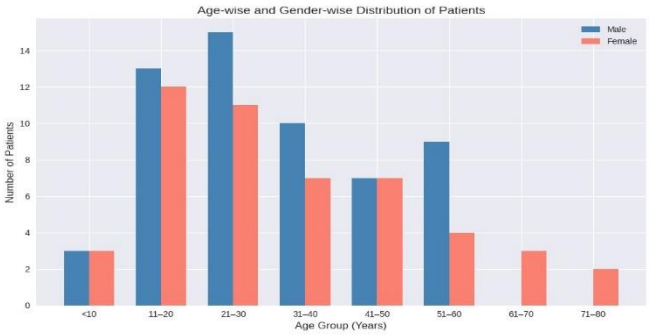

Figure 1. Shows the highest distribution of OKC in the third and second decade with a slight male predominance.

At the time of initial presentation, the patients demonstrated diverse clinical symptoms including pain, swelling, ulceration and discharge. (Figure 2).

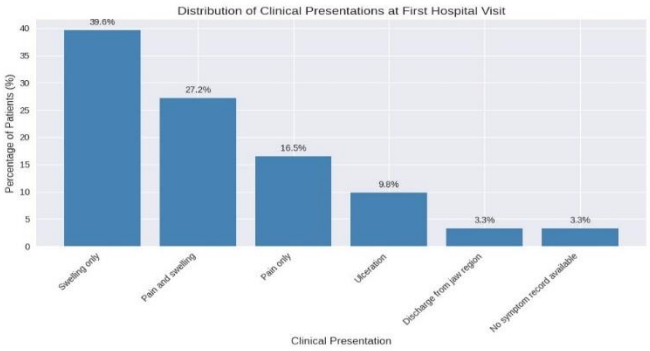

Figure 2. Shows the common symptoms of the patients at their first visit to the hospital.

Among 120 patients with OKC, 106 individuals were diagnosed with having sporadic odontogenic keratocysts. The radiographic details revealed that 73% of OKC patients presented with radiolucent lesions. The distribution of radiographic presentations is illustrated in figure 3, which shows unilocular radiolucency as the common radiographic finding.

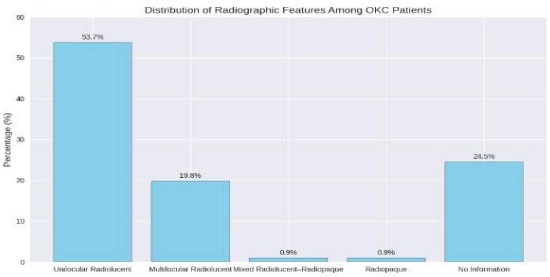

Figure 3. Shows various radiographic presentation among OKC patients.

The 14 patients who had multiple cystic lesions in various parts of the jaws showed presence of multiple radiolucent lesions. The details of these patients were

assessed and were categorized as syndromic (Gorlin-Goltz syndrome) or not based on information available for the diagnosis of the syndrome. Among 14 patients, only eight patients met with the diagnostic criteria for Gorlin-Goltz syndrome. Additionally, six patients presented with multiple OKCs and clinical signs suggestive of systemic involvement; however, they did not fulfil the complete diagnostic criteria for Gorlin-Goltz syndrome.

Excluding patients diagnosed with Gorlin-Goltz syndrome and those presenting with other systemic involvement, the mandibular posterior region emerged as the most frequent site of occurrence for odontogenic keratocysts followed by the maxillary posterior region, maxillary anterior region and mandibular anterior region. 13.1% of lesions crossed midline and the details of 1.8% was not mentioned (Figure 4).

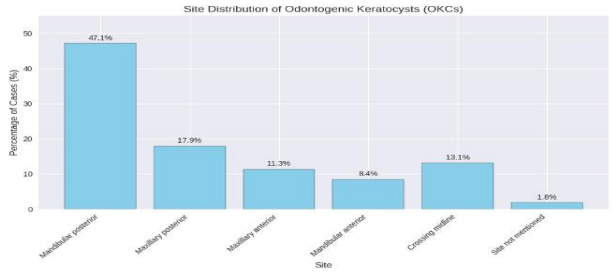

Figure 4. Shows the mandibular posteriors as the common site for OKC followed by the maxillary posterior region. Among the maxillary region, the anterior teeth are commonly affected compared to posterior teeth.

An association with impacted teeth was observed in 28% of OKC cases, with the third molars being the most commonly involved, followed by canines, second premolars, and lower incisors (figure 5). In patients presenting with multiple jaw lesions, OKCs were predominantly located in the mandibular posterior region with additional involvement noted in the maxillary and mandibular anterior regions.

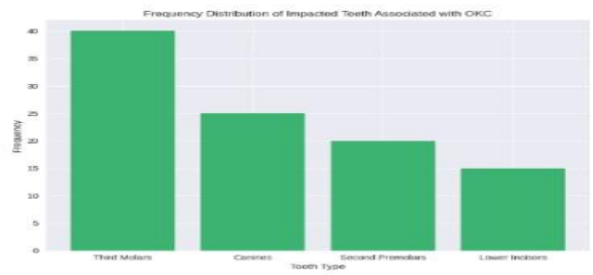

Figure 5. Shows distribution of impacted teeth associated with odontogenic keratocysts.

Among patients with odontogenic keratocysts, 8 individuals (6.6%) demonstrated clinical features consistent with Gorlin-Goltz syndrome. The age range of these patients spanned from 8 to 36 years with a mean age of 23 years. Among these, a marked male predominance was seen comprising of 7 males and one female. All the findings documented in the registry were collected and the major and minor criteria were considered. On evaluating the patients for Gorlin-Goltz syndrome, multiple odontogenic keratocysts was found in all patients. The distribution of the jaw lesions among 8 patients with Gorlin-Goltz are illustrated in figure 6.

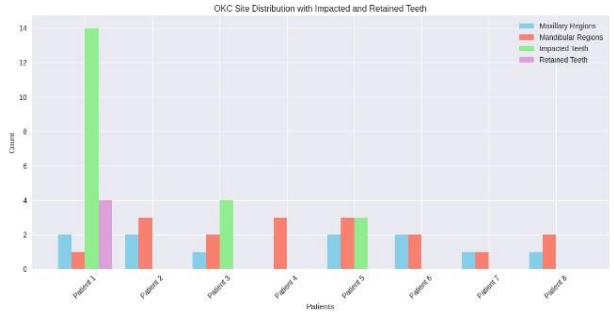

Figure 6. Shows the highest distribution of multiple OKC in the mandibular region compared to maxillary region.

Other major criteria found along with multiple OKC's were calcification of the falx cerebri, palmar and plantar pitting and bifid ribs. A positive family history was noted in one patient, whose brother and father were also diagnosed with Gorlin-Goltz syndrome. Among the minor diagnostic criteria, the most common findings include the ocular anomalies, hypertelorism, cleft lip or palate, polydactyly and syndactyly, frontal bone defects, scoliosis or kyphosis, vertebral fusion, hypoplastic maxilla, tendinous calcification and a depressed nasal bridge (figure 7).

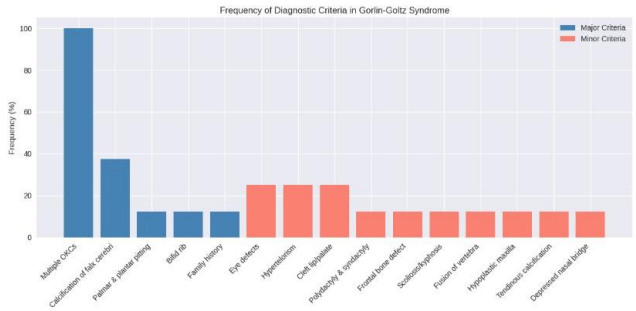

Figure 7. Shows the distribution of major and minor criteria among patients with Gorlin- Goltz syndrome.

An additional six patients presented with multiple OKCs with signs of systemic involvement.

However, they did not meet the diagnostic criteria for Gorlin-Goltz syndrome due to incomplete documentation. The details of these patients were recorded separately.

DISCUSSION

Gorlin–Goltz syndrome, also referred to as Gorlin syndrome or nevoid basal cell carcinoma syndrome (NBCCS), is a rare autosomal dominant disorder characterized by variable penetrance and expressivity[10,15,23,28]. The earliest documentation of this syndrome can be traced back to 1864, when Jarish and White initially described its characteristic features. However, it was in 1960 that the condition was formally named Goltz-Gorlin syndrome[8]. Though nevoid basal cell carcinoma syndrome is primarily associated with mutations in the PTCH1 gene, it has also explored the role of SUFU, a key repressor of the sonic hedgehog (SHH) signalling pathway, as a contributing genetic factor[10,13,29]. Several predisposing factors like excessive sun exposure, radiation, immunosuppression and vitamin D deficiency have been implicated in accelerating disease presentation and worsening prognosis[13,29].

Though the initial diagnosis of this syndrome was established by Evans et al based on a characteristic triad of symptoms, modifications have been done over the years by Kimonis et al and Bree et al[15,16]. This syndrome presents with a wide spectrum of systemic manifestations, including developmental anomalies and neoplastic growths. Due to its variable expressivity and multisystem involvement, diagnosis relies heavily on clinical presentation. The established diagnostic criteria require one of the following combinations for the diagnosis of the syndrome: (1) a single major criterion with molecular confirmation, (2) two major criteria, or (3) one major and two minor criteria [6-10,15]. One of the significant findings noticed in this syndrome is its association with OKC, which is one of the earlier manifestations of the syndrome. Therefore, the present study was focused on the association of OKC with Gorlin-Goltz syndrome.

Demographic Profile

The male-to-female ratio of 1.2:1 observed in this study indicates a slight male predominance, consistent with several reports in the literature[3,5,30,31,33]. However, the youngest and oldest patients, aged 11 months and 72 years respectively were both females, highlighting the wide age spectrum of OKC occurrence. The mean age of 40 years with a peak incidence in the third decade followed by the second decade aligns with previous studies that suggest OKCs are most common in young adults[32,33].

Site Distribution and Tooth Association

The mandibular posterior region was the most common site of occurrence followed by the maxillary posterior region. This distribution is consistent with the predilection of OKCs for the posterior mandible reported in earlier studies[3-5,12,32]. Association with impacted teeth was observed in 28% of cases, predominantly involving third molars, which corroborates the established relation between OKCs and unerupted teeth[12,33,35].

Clinical Presentation

Patients presented with a variety of symptoms ranging from swelling, pain, drainage and ulceration which is similar to previous literature[4,5]. This could be due to the fact that OKCs often remain asymptomatic until they reach a considerable size, when the swelling becomes the predominant complaint. The presence of ulceration or drainage might reflect secondary infection or cortical perforation. Notably, documentation gaps in 3.3% of cases emphasize the importance of meticulous clinical record-keeping for accurate epidemiological assessment.

Radiographic Features

Radiographic evaluation revealed that unilocular radiolucency as the most common pattern, while mixed radiolucent–radiopaque and purely radiopaque presentations were rare[32-35]. However, the absence of radiographic data in nearly one-fourth of cases underscores the need for standardized imaging protocols in diagnostic work-up.

Syndromic Association:

Of the 120 patients with OKC, 6.6% exhibited features consistent with Gorlin–Goltz syndrome, aligning with the findings reported by Brannon and Myoung et al[32,35]. The mean age of syndromic patients was 23 years, with a notable male predominance (7 males, 1 female). While the mean age matched earlier studies, such a distinct male predominance has not been documented in the literature. The most consistent major criterion was the presence of multiple odontogenic keratocysts observed in all syndromic patients (100%). Additional major features included calcification of the falx cerebri, palmar/plantar pits, and bifid ribs. Minor criteria comprised ocular anomalies, hypertelorism, cleft lip/palate and skeletal abnormalities. These findings were generally in line with previously published reports, though variations in the percentage distribution among affected individuals were noted[6,7,15,17,18,20,23]. A significant difference in this study compared to earlier reports was the absence of basal cell carcinoma and other neoplastic lesions. When compared with studies on the Indian population, the results were largely similar, except for a higher incidence of cleft lip/palate and scoliosis, which was rarely reported in other groups[19,22,25-27]. Interestingly, only one case series documented basal cell carcinoma, whereas the other Indian case series have not been reported this condition. The absence of extensive studies pertaining to Gorlin-

Goltz syndrome suggests that the syndrome is either rare in the Indian population or may be under-reported owing to lack of awareness about the clinical and radiological manifestations of Gorlin-Goltz syndrome.

These observations underscore the importance of comprehensive systemic evaluation in patients presenting with multiple OKCs. Early recognition of Gorlin–Goltz syndrome is crucial for effective multidisciplinary management and genetic counselling. The findings also highlight the aggressive nature of OKCs, their tendency to recur and their association with syndromic conditions. Given the variability in clinical and radiographic presentations, clinicians must maintain a high index of suspicion. Moreover, the identification of Gorlin–Goltz syndrome in a subset of patients emphasizes the need for close collaboration among oral pathologists, radiologists, dermatologists, and geneticists to ensure holistic patient care.

This being the first study to assess the prevalence of Golin Goltz syndrome among OKC in Indian population, there are certain limitations which should be considered. First, the retrospective design relied only on the available clinical records, which may have led to incomplete documentation of syndromic features. Second, the sample was drawn from a single institution, which may limit the generalizability of the findings to broader populations. Third, genetic testing was not performed and the diagnosis of Gorlin-Goltz syndrome was based solely on clinical and radiographic criteria, which could underestimate or overestimate true prevalence. Finally, the relatively small number of patients identified with Gorlin-Goltz syndrome restricts the ability to draw definitive conclusions regarding demographic variability. Future prospective, multicentric studies incorporating molecular confirmation are recommended to validate and expand upon these findings.

CONCLUSION

This retrospective study highlights the association between odontogenic keratocysts (OKCs) and Gorlin-Goltz syndrome in South Indian population. The findings suggest that the expression of diagnostic criteria for Gorlin-Goltz syndrome may vary across demographic groups, emphasizing the need for population-specific studies and heightened clinical vigilance. Early recognition of the syndrome is crucial for comprehensive patient management, given its multisystem involvement and potential complications. Future multicentric studies with larger cohorts are warranted to better understand the prevalence, variability, and clinical implications of Gorlin-Goltz syndrome in diverse populations.

DECLARATIONS

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Competing Interests

The authors have no competing interests to declare.

Informed Consent

Not applicable.

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