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Gorlin-Goltz Syndrome

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Gorlin-Goltz Syndrome (GGS) is known as nevoid basal cell carcinoma syndrome (NBCCS); it is a rare condition with multi-organ involvement. It has an autosomal dominant trait with complete penetrance and variable expressivity. The condition presents with a wide range of pathological features including malignancy of the skin (basal cell carcinoma); its early diagnosis is vital. Odontogenic Keratocyst (OKC) being one of the prominent features and mostly an early one, the dental specialty most often is in a position to identify this condition first.

We present a case of GGS in a fourteen-year-old male. Enucleation was performed and the postoperative period was uneventful. Histopathologically, the diagnosis was confirmed as multiple Odontogenic Keratocyst. The patient had spina bifida at D2 vertebra, fused anterior end of right 5th and 6th ribs, fused right anterior 2nd and 3rd ribs, Falx and tentorial calcification and Sprengel shoulder. It is essential to emphasize the role of the dental specialty in diagnosing and instituting early treatment of such condition.

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Gorlin-Goltz Syndrome (GGS) is a rare genetic condition, which has an autosomal dominant inheritance with variable expressiveness. Though it was first reported by Jarish as early as 1894, its recognition as a syndrome became established by the works of Gorlin in 1960[1,2]. Earlier to Gorlin, it had been known by a multitude of names that compounded the confusion. Its unique importance as a clinical entity stems from its multi-organ involvement with the potential of skin malignancies[3].

A wide variety of clinical and radiological features of GGS have been described. OKC is known as Keratocystic Odontogenic tumor (KOT)[4]. The management of KOT is essentially enucleation and regular long-term follow-up because of its high recurrence rate.

The aim of this presentation is to report a rare case of Gorlin-Goltz syndrome, which was managed by enucleation and regular follow-up.

THE CASE

A fourteen-year-old male complained of pain and swelling of lower left quadrant of the jaw. Medical history revealed similar episode three months earlier which settled with a course of antibiotics. The patient appeared weak, skinny and physically inactive for his age. On examination, he had a fluctuant and tender swelling 3x3 cm involving lower left premolars. There was pus discharging along the gingival sulcus. A clinical diagnosis of an acute dentoalveolar abscess was made, drainage was done, culture and sensitivity was requested and he was empirically placed on Augmentin. Orthopantomograph and CT revealed multiple well-circumscribed radiolucent lesions, five in number, three on the maxilla and two in the mandible, see figures 1, 2 and 3. All these lesions appear to be associated with an impacted or unerupted tooth. Though multiple dentigerous cysts are extremely rare, radiological diagnosis confirmed that.

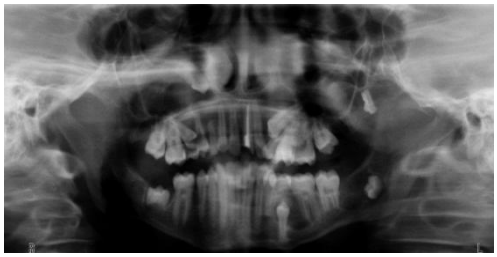
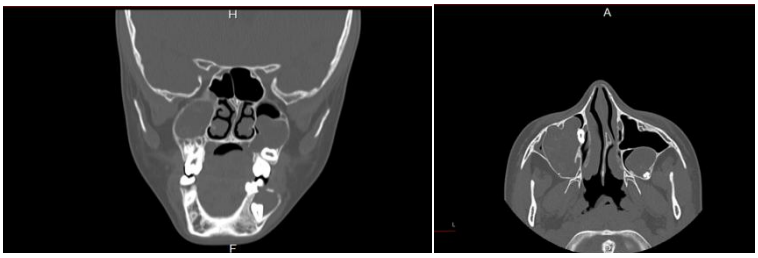


Figure 1: Orthopantogram Showing Cystic Lesions in the Mandible and Maxilla Associated with Teeth



Figures 2 and 3: CT Scan of the Maxilla and Mandible Showing Multiple Well-Circumscribed Radiolucent Lesions

Enucleation was performed after the acute condition settled, the postoperative period was uneventful.

Histopathologically, the diagnosis was confirmed as multiple Odontogenic Keratocyst, known as KOT, see figure 4. CT brain, x-ray skull and x-ray chest showed spina bifida at D2 vertebra, fused anterior end of right 5^{th} and 6^{th} ribs, fused right anterior 2^{nd} and 3^{rd} ribs fused, Falx and

tentorial calcification and Sprengel shoulder, see figure 5. The patient confirmed to have two major and three minor criteria.

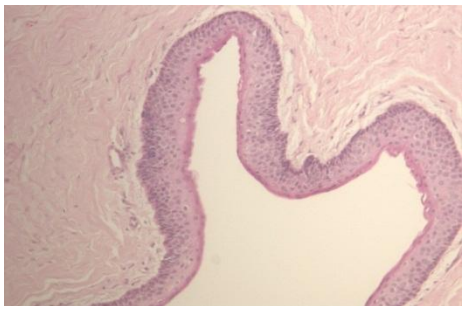


Figure 4: Histopathology Showing Uniform Keratinized Epithelium with Palisading of the Basal Cell Layer

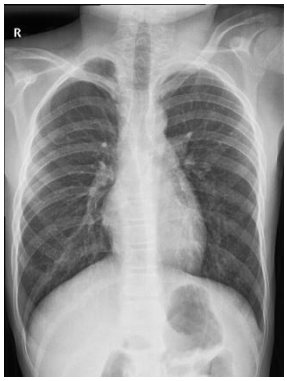


Figure 5: Chest X-Ray Showing Presence of Bifid and Fused Ribs

DISCUSSION

GGS is a condition that demands an early diagnosis because of its multiple malignant potentials. The prevalence of this condition is one in 60,000[5]. There has been no sexual predilection and mostly it arises in the first, second or third decade[6]. The features of GGS include basal cell carcinoma, multiple odontogenic keratocyst, skeletal abnormalities, ectopic calcification, palmar and plantar pits[7]. The major criteria are the following: multiple basal cell carcinomas or one occurring under the age of 20 years, histologically proven OKCs of the jaws, palmar or plantar pits (three or more), lamellar calcification of the falx cerebri and bifid fused or markedly splayed ribs. The minor criteria are the following: macrocephaly, congenital malformation, such as cleft lip or palate, frontal bossing, coarse face, moderate or severe hypertelorism, other skeletal abnormalities: Sprengel deformity, marked pectus deformity, marked syndactyly of the digits, bridging of the sella turcica, vertebral anomalies such as hemi vertebrae, fusion or elongation of the vertebral bodies, modeling defects of the hands and feet or flame shaped hands or feet, Ovarian fibroma and Medulloblastoma.

Our case revealed three major and two minor criteria. The skin lesion, though absent in the reported case, it should be looked for because of the malignant potential (50%-97%). As OKC is

considered one of the major criteria and occasionally a feature that presents initially, the dental specialty is placed in the forefront for the diagnosis[9].

The WHO reclassified OKC to KOT because of its aggressive nature and high-rate of recurrence. This lesion has been reported to occur in at least 90% of the patients with GGS, mostly the mandible, 44% occur in the mandibular angle. OKC is relatively less frequent in the maxilla, but tend to be more aggressive than those in the lower jaws. The KOTs are divided into parakeratotic, orthokeratotic and rarely mixed. The cavity is filled with thick keratinous material or a straw-colored liquid. The radiographic images show well-circumscribed radiolucency, mostly unilocular in the initial stages and may develop into multilocular. Expansion of the bones and asymmetry may be a feature though not always. Inflammatory symptoms of the jaw lesions sometimes encourage patients to seek medical help and hence an early detection[9]. Occlusal and dental anomalies are mostly a regular feature in patients with multiple OKC. The associated teeth may be displaced or its eruption impaired to give it a radiological diagnosis of dentigerous cyst.

Treatment of KOT depends on various factors including age, extent and location of the lesion. Basically, the treatment is enucleation or resection. However, the choice of treatment method would depend on patient's age, the functional and aesthetic concerns. The consensus is to perform an enucleation followed by chemical curettage using Carnoy's solution to prevent recurrence[10].

Approximately 60% of GGS patients may have a recurrence of OKCs. However, these patients show tendencies to develop new cysts[11]. It has been shown that recurrence of OKCs in patients with GGS would be higher than OKCs in otherwise healthy patients.

CONCLUSION

A patient diagnosed with Gorlin-Goltz syndrome was presented. The role of dental specialty in its diagnosis was emphasized as KOT is a prominent feature of this syndrome. Early diagnosis and regular long-term follow-up are strongly recommended.

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REFERENCES

1. Jarish W. Zur Lehre Von Den Autgeschwulsten. Archive Jur Dermatologic and Syphilogic 1894; (28):163-222.
2. Gorlin RJ, Goltz RW. Multiple Nevoid Basal-Cell Epithelioma, Jaw Cysts and Bifid Rib. A Syndrome N Engl J Med. 1960; 262:908-12.
3. Ortega García de Amezaga A, García Arregui O, Zepeda Nuño S, et al. Gorlin–Goltz Syndrome; Clinicopathologic Aspects. Med Oral Patol Oral Cir Bucal 2008; 13(6):E338-43.
4. Madras J, Lapointe H. Keratocystic Odontogenic Tumour: Reclassification of the Odontogenic Keratocyst from Cyst to Tumour. J Can Dent Assoc 2008; 74(2):165-165h.
5. Gorlin RJ. Naevoid Basal Cell Carcinoma (Gorlin) Syndrome: Unanswered Issues. J Lab Clin Med 1999; 134(6):551-2.
6. Manfredi M, Vescovi P, Bonanini M, et al. Nevoid Basal Cell Carcinoma Syndrome: A Review of the Literature. Int J Oral Maxillofac Surg 2004; 33(2):117-24.
7. Casaroto AR, Loures DC, Moreschi E, et al. Early diagnosis of Gorlin-Goltz Syndrome: Case Report. Head Face Med. 2011; 7:2.
8. Evans DG, Ladusans EJ, Rimmer S, et al. Complications of the Naevoid Basal Cell Carcinoma Syndrome: Results of a Population Based Study. J Med Genet 1993; 30(6):460-4.
9. Ahn SG, Lim YS, Kim DK, et al. Nevoid Basal Cell Carcinoma Syndrome: A Retrospective Analysis of 33 Affected Korean Individuals. Int J Oral Maxillofac Surg 2004; 33(5):458-62.
10. Blanchard SB. Odontogenic Keratocysts: Review of the Literature and Report of a Case. J Periodontol 1997; 68(3):306-11.
11. Lo Muzio L, Nocini P, Bucci P, et al. Early Diagnosis of Nevoid Basal Cell Carcinoma Syndrome. J Am Dent Assoc 1999; 130(5):669-74.