Case Report DOI: 10.22114/ajem.v0i0.83

A Rare Case of Gorlin-Goltz Syndrome Presented to the Emergency Department as Facial Swelling

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Published online: 2018-04-21

Abstract

Introduction: Gorlin-Goltz syndrome (GGS), also known as basal cell nevus syndrome, is a very rare autosomal dominant inherited disorder that is characterized by the development of numerous basal cell carcinoma. This article reports a case of GGS, emphasizing its clinical and radiographic manifestations.

Case presentation: We report here the case of a 35-year-old man who visited the maxillofacial emergency department due to left facial swelling. According to his clinical and radiographic examination we diagnosed him with GGS with no family history. The patient has multiple odontogenic keratocysts, rib anomalies, calcifications of the falx cerebri, lower jaw prognathism, frontal bossing, macrocephaly, and thick eyebrows.

Conclusion: A definitive diagnosis of GGS should be made by a multidisciplinary team including a maxillofacial surgeon and medical specialists. Early diagnosis, treatment, and regular follow up are important to decrease complications, including oromaxillofacial deformation and destruction, and possible malignancy.

Key words: Basal cell nevus syndrome; Case reports; Odontogenic cysts; Oral and maxillofacial surgeons

Cite this article as: Aloosi SN, Mahmood KA, Ali ShM, Mahmud PKh, Hasan SO, Muhamed HO. A Rare Case of Gorlin-Goltz Syndrome Presented to the Emergency Department as Facial Swelling. Adv J Emerg Med. 2018;2(4): e46.

INTRODUCTION

Gorlin-Goltz syndrome (GGS), also known as basal cell nevus syndrome, is a very rare autosomal dominant inherited disorder that is characterized by the development of numerous basal cell carcinomas (1). It is presented mainly by the presence of odontogenic keratocysts (OKCs) of the jaw and palmar pits and is also associated with a wide spectrum of developmental anomalies and neoplasms. This article reports a case of GGS, emphasizing its clinical and radiographic manifestations.

CASE PRESENTATION

A 35-year-old man visited the emergency department (ED) of Sulaimany Hospital for left facial swelling of 2 months duration that was gradually increasing in size, and was recently causing pain.

The patient was examined by a maxillofacial surgeon physician for further history taking and physical examination. His past medical history was not significant. Extraoral examination revealed macrocephaly, frontal bossing, hypertelorism, a wide nasal bridge and thick eyebrows. A diffuse swelling was present in the left mandibular mental nerve region that was firm to hard in consistency and was mildly tender on palpation (figure 1). The skin of his palms and soles showing hyperkeratosis and brownish pinpoint papules in the neck, chest, and forearm. Intraoral examination revealed malocclusion and

Figure 1: Photograph showing frontal boosing, hypertelorism, wide nasal bridge, prognathism, and thick eyebrows with swelling in the left mandibular region

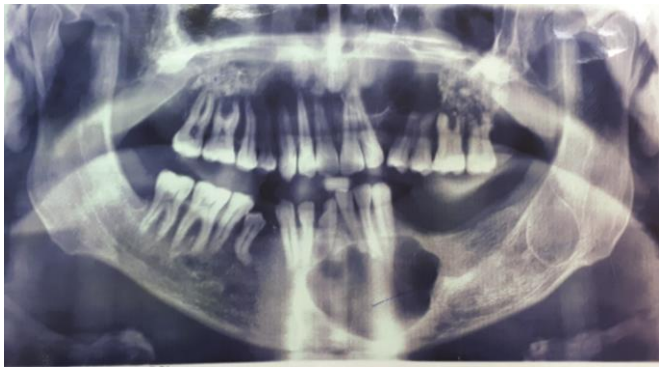

Figure 2: Orthopantomogram shows multiple odontogenic keratocysts in both maxilla and mandible

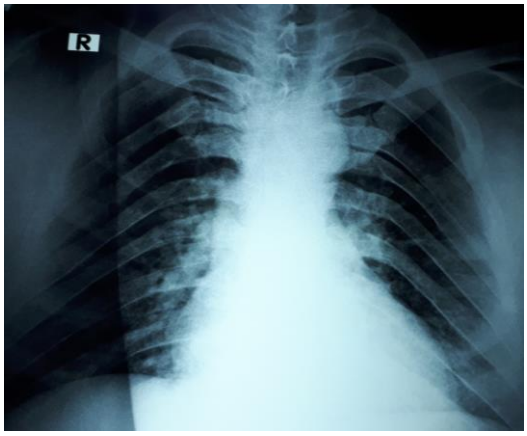

Figure 3: Chest x-ray revealed a bifid rib

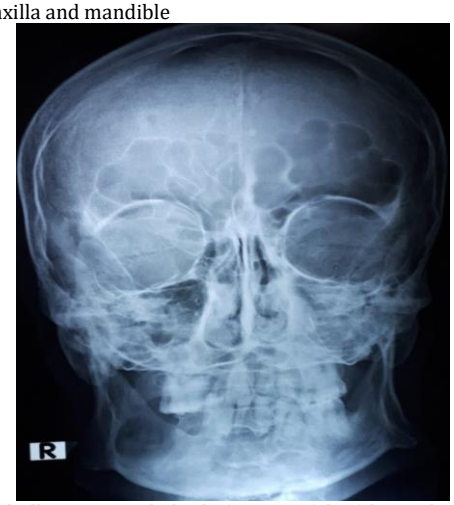

Figure 4: Skull x-ray revealed calcification of the falx cerebri

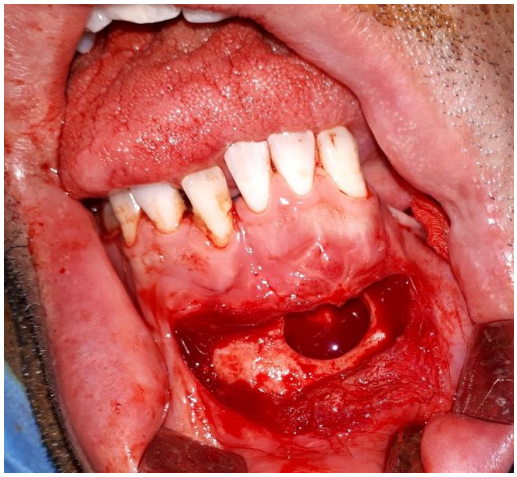

Figure 5: Photograph showing surgical enucleation of the cyst that was sent for histopathological examination.

a partially edentulous situation with diffuse swelling in the left mandibular mental nerve region and the left maxillary canine region. The overlying mucosa was firm to hard in consistency and was tender on palpation.

Radiographic investigations including orthopantomogram (OPG) of the patient showed multiple cysts (radiolucencies) present in both the maxilla and mandible of the patient (figure 2). A radiograph of the skull showed calcification of the falx cerebri (figure 3) and bifid ribs are apparent on his chest radiograph (figure 4). Based on the clinical history and radiographic findings, the case was provisionally diagnosed as GGS.

Blood investigations were within the normal limits and therefore surgical enucleation of the cystic lesions was performed (figure 5). The histopathology of the excised epithelium revealed the presence of stratified squamous epithelium with para keratinization overlying the connective tissue, thereby confirming the diagnosis of a keratocystic odontogenic tumor (figure 6). Therefore, based on clinical features, imaging characteristics, and histological findings a final diagnosis of GGS was made.

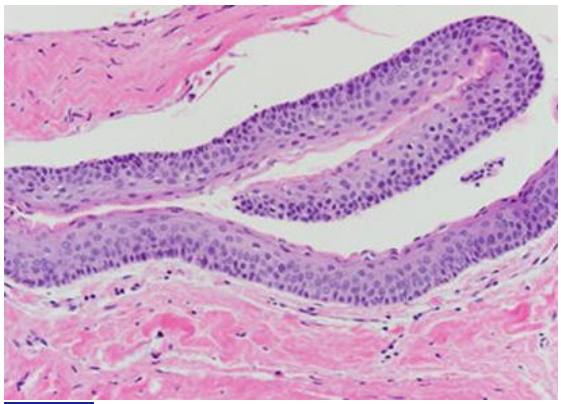

Figure 6: Histopathology findings revealed the presence of stratified squamous epithelium with para keratinization overlying the connective tissue

DISCUSSION

GGS was described by Jarisch and White for the first time in 1894. The disease is an autosomal dominant disorder and its prevalence is estimated to be about 1 in 60,000. It is seen equally in men and women. It is caused by mutations in PTCH, a tumor suppressor gene that is a human homologue of the Drosophila segment polarity gene. PTCH located on the long arm of human chromosome 9q22.3 (2-5).

It is mainly characterized by the presence of multiple basal cell carcinomas, odontogenic keratocysts of the jaw and palmar pits and is also associated with a wide spectrum of developmental anomalies and neoplasms, hypertelorism, calcification of the falx cerebri, palmer and planter pits, central nervous system defects, ocular lesions, rib anomalies, mandibular prognathism (class III jaw relationship), cleft lip, and palate. Other findings of this syndrome in rare cases include ovarian fibromas (6, 7). The oral and maxillofacial surgeon has the main role in diagnosing the syndrome because multiple odontogenic keratocysts are the first manifestations of this syndrome (8).

Evans et al. in 1994 first established major and minor criteria for the diagnosis of this rare entity, which were modified later by Kimonos et al. in 2004 (9, 10). Currently, 2 major criteria or 1 major and 2 minor criteria are necessary in order to diagnose GGS (11). The major and minor criteria are as follows:

Major criteria

More than two basal cell carcinomas or one in a patient less than 20 years old; Odontogenic keratocysts of the jaw; Bilamellar calcifications of the falxcerebri and tentorium; Three or more palmar or plantar pits; Bifid, fused, or markedly splayed ribs; First-degree relative with GGS

Minor criteria

Macrocephaly; Congenital anomalies (cleft lip or palate, frontal bossing, coarse facies, and moderate or severe hypertelorism); Other skeletal anomalies (Sprengel deformity, marked pectus deformity, and marked syndactyly of the digits); Radiologic anomalies (such as bridging of the sellaturcica, vertebral anomalies, modeling defects of the hands and feet, or flame-shaped lucencies of the hands and the feet); Medulloblastoma, seizures, mental retardation, meningioma; Ovarian fibroma

In our case, three major criteria—odontogenic keratocysts of the jaw, calcification of the falx cerebri and a bifid rib in the right side of chest along with two minor criteria—macrocephaly, frontal bossing, and moderate hypertelorism were present. Other recognizable features of the syndrome were prognathism, palmer pits, a solitary pigmented nevus (mole), and areas of hyperpigmentation in the neck and forearm confirmed the diagnosis as GGS. The treatment of GGS is in accordance with the generally accepted rules for the treatment of basal cell carcinomas and keratocysts in other patients. Radiation should be avoided because it may trigger the development of additional tumors in the adjacent skin areas.

CONCLUSIONS

Every maxillofacial surgeon or dentist, especially oral radiologists, pedodontists, and orthodontists, have an important responsibility in the early diagnosis of this condition. A definitive diagnosis of GGS also should be made by a multidisciplinary team that includes a maxillofacial surgeon and medical specialists. Early diagnosis, treatment, and regular follow up are important to decrease complications, including oromaxillofacial deformation and destruction and the possibility of malignancy.

ACKNOWLEDGMENTS

Here in Iraq, we do not have committees or organizations for ethical approval. Taking informed consent is the only standard ethical process within hospital permission rules because all hospitals in Iraq are teaching governmental hospitals and therefore informed consent is taken before doing any procedure or publishing the information of any patient. Written informed consent was obtained from the patient for publication of this case report and the accompanying images.

AUTHORS' CONTRIBUTION

SNA performed the surgery; SMA designed and wrote the manuscript and participated in the operation, KAM performed the radiological investigation, PKHM and HOM received and followed up the patient, and all authors read and approved the final manuscript.

CONFLICTS OF INTEREST

All authors of this manuscript declare that they have no conflicts of interest with any person or organization.

FUNDING

This work was performed solely by the authors of this manuscript. No one else participated in the

preparation of or provided financial aid for this work. The examinations and treatment were performed in a public hospital that is open to everybody. We just used extra time to collect the data and put it together in the form of this case report.

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