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Case Report of Gorlin-Goltz Syndrome

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Abstract **Case Report**

Gorlin-Goltz syndrome, also known as basal cell nevus syndrome. This condition is characterized by various clinical manifestations, including multiple cutaneous lesions and other systemic abnormalities. We present the case of a 32-year-old man with no significant medical history who was referred to our institution for evaluation of multiple budding lesions. Clinical examination revealed three lesions in the upper right nasolabial fold, one in the lower right cheek, one in the left nasolabial fold, and additional suspect lesions on the chin, right temple, and right cheek. This case underscores the importance of recognizing the diverse manifestations of Gorlin-Goltz syndrome, which can aid in early diagnosis and management of this inherited disorder. Further investigation and long-term follow-up are essential for understanding the progression and management of this condition.

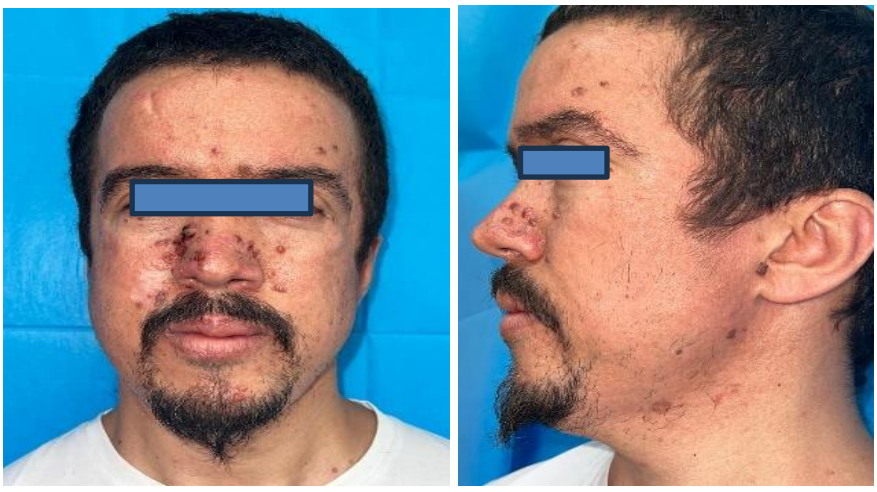
Keywords: Gorlin-Goltz syndrome, basal cell nevus syndrome, neurocristopathy, keratocyst.

I. INTRODUCTION

Gorlin-Goltz syndrome, also known under various names such as basal cell nevus syndrome, the fifth phakomatosis, Ward syndrome, or generalized multiple epitheliomatosis, is currently classified as a neurocristopathy. It is an inherited syndromic dysembryoplastic disorder.

II. CLINICAL OBSERVATION

A 32 YEAR old man without a significant medical history was referred to our institution because of a multiple budding lesions: Three in the upper right nasolabial fold, one in the lower right cheek, one in the left nasolabial fold and suspect lesions: chin, right temple, right cheek and two bilateral vestibular swellings covered with intact mucosa. On palpation, the swellings are painless, firm but depressible, with no dental mobility. The general examination reveals palmar and plantar pits **Figure 1**.



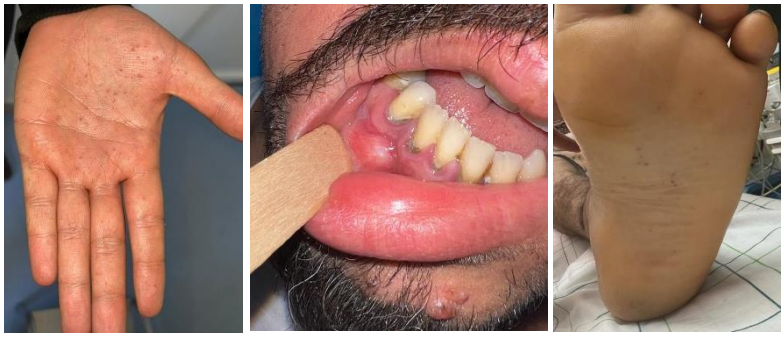


Figure 1: Oral and extra oral clinical manifestation

III. Diagnostic Assessment:

A facial CT scan was performed, revealing: Lytic mandibular masses in the symphyseal and right parasymphyseal areas, Swelling of the cortical bone, bilateral involvement of the inferior alveolar canal, suggesting mandibular keratocysts. **Figure 2**. Biopsies of the suspect skin lesions revealed nodular basal cell carcinomas (BCC), while biopsies of the mandibular cystic lesions confirmed mandibular keratocysts.

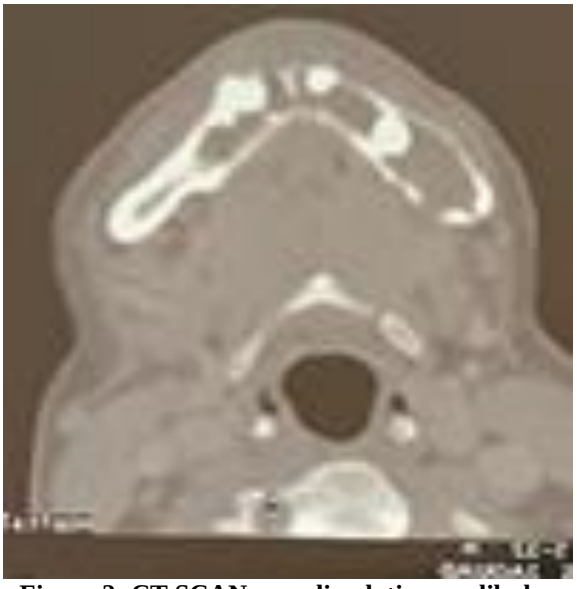


Figure 2: CT SCAN revealing lytic mandibular masses

The diagnosis of Gorlin-Goltz syndrome is based on the following criteria:

- Multiple BCCs, Mandibular keratocysts, cerebral calcifications and palmar and plantar pits.
- The patient was operated under general anesthesia, the procedure consisted in a first stage of an excision of the upper right nasolabial, lower right nasolabial and left nasolabial Basal Cell cancer, with respect for 5mm excision margins.
- Excision of a suspicious right chin, temporal and cheek lesion with a 5mm margin
- The Reconstruction of the right nasolabial fold using a Mustarde flap **Figure 2.**
- An enucleation of 3 mandibular cysts was carried out associated with curettage **Figure 3.**

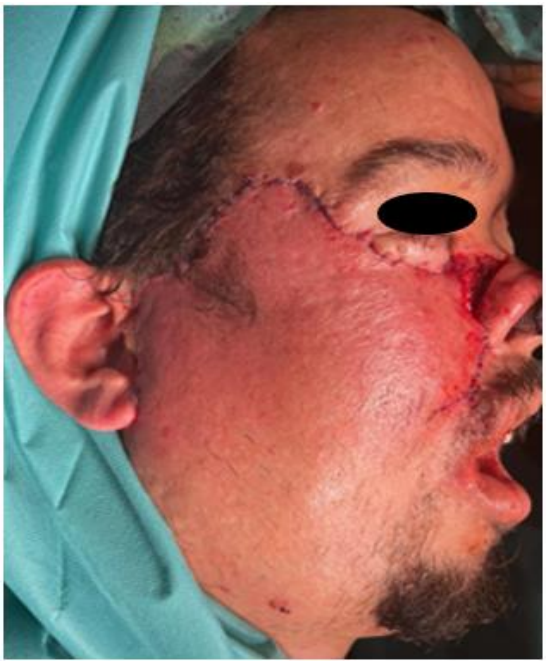


Figure 3: Intraoperative Image: Reconstruction using Mustarde FLAP

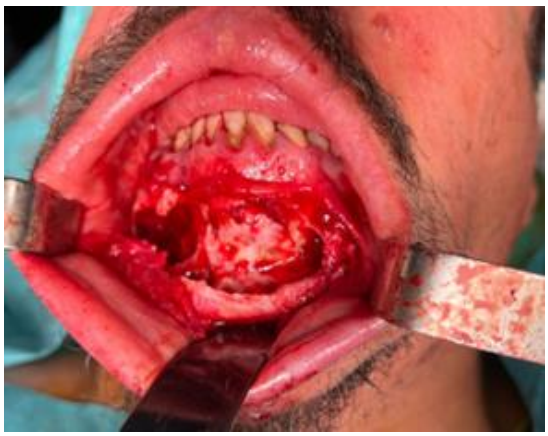


Figure 4: Intraoperative Image: Curettage of three mandibular cysts

IV. DISCUSSION

Gorlin-Goltz syndrome or nevoid basal cell carcinoma syndrome (NBCCS) was first described in

1894 by Jarish and White, but better defined by Gorlin and Goltz in 1960 [1,2]. It is a rare disorder due to mutation in the patched tumor suppressor gene PTCH, located in chromosome 9q22.3q31, which functions as a component of the Hedgehog signaling pathway. Clinical manifestations are diverse: cutaneous, skeletal, ocular, neurological, genital, and renal [3,4]. Incidence of basal cell carcinomas in Gorlin-Goltz syndrome varies among ethnic groups and is reported in up to 90% of affected Caucasians [5].

The diagnosis is based on clinical findings and confirmed by the presence of two major criteria or one major associated with two minor ones: The major criteria are: more than 2 Basal cell carcinomas or 1 before 20 years of age; odontogenic keratocysts confirmed by histology; one or more palmoplantar pits; bilamellar calcification of cerebral falx; fused or flattened bifid ribs; 1st degree relative affected. The minor criteria are: macrocephaly; congenital malformations (cleft lip or palate, frontal bossing, coarse facies, hypertelorism); skeletal alterations (Sprengel deformity, deformed chest, hemivertebrae, fusion or lengthening of vertebral bodies, anomalies in hands and feet, syndactyly, candle-flame shaped hand bone ysts); pointed sella turcica; ovarian fibroma; medulloblastoma [6].

In view of the wide clinical spectrum of this syndrome, the management of its modalities is not standardized Recommendations for patients include: UV Protection using of high SPF sunscreen to prevent BCCs Treatment of Keratocysts by conservative methods (enucleation) or aggressive methods (block resection).

Treatment of BCCs by Oncological approach. As a general rule, radiotherapy is avoided due to the intense sensitivity of these individuals to ionizing radiation. The best conduct is exeresis and, for extensive areas with many BCCs, photodynamic therapy with 5-aminolevulinic acid is an option [7].

V. Declaration of Interests

The authors declare no conflicts of interest related to this case report.

VI. CONCLUSION

Gorlin-Goltz syndrome is rare but not exceptional. It frequently combines basal cell nevus syndrome, jaw keratocysts, and cerebral calcifications. The severity of the disease, due to its carcinogenic potential, justifies early screening and regular monitoring.

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