Gorlin-Goltz Syndrome with *PTCH1* Gene Mutation in a Family Presenting with Odontogenic Keratocysts

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A 19-year-old female presented with mandibular swelling and mild spontaneous pain. Panoramic radiography revealed cystic lesions extending from the right mandibular ramus to tooth 36, as well as an additional lesion at tooth 28 (Figure 1a). Cone-beam computed tomography (CBCT) showed multilocular radiolucencies with cortical destruction and internal septa in the same regions (Figure 1b). Enucleation was performed for the lesions near teeth 48 and 28. Histopathology confirmed that odontogenic keratocysts (OKCs) lined with parakeratinized epithelium. Due to its size and involvement with multiple teeth, the anterior mandibular lesion was managed with marsupialization to preserve tooth vitality. After approximately 1 year of treatment, the lesions resolved uneventfully. The patient has been monitored regularly for 23 years, with no recurrence to date.

At the 11-year follow-up, the patient's 12-year-old son presented with asymptomatic swelling. Clinical examination revealed facial asymmetry and a painless intraoral mass. Panoramic radiography showed a large cystic lesion surrounding the mandibular canine and multiple cysts in all four quadrants (Figure 1c). CBCT confirmed the size and expansion of these lesions (Figure 1d). Marsupialization was performed for the right posterior mandibular cyst. Histopathology again confirmed OKCs. Numerous cutaneous nevi were noted, and dermatologic referral with excisional biopsy identified a basal cell carcinoma (BCC) in one nodular lesion. Genetic testing revealed *PTCH1* gene mutations in both the patient and his mother. Given the presence of multiple OKCs, a first-degree relative with Gorlin-Goltz syndrome (GGS), and confirmed BCC, the diagnosis of GGS was established in 2014. The anterior mandibular lesion healed with preservation of tooth vitality. The remaining cysts were enucleated. At the 12-year postoperative follow-up, a recurrence in the region of tooth 48 was detected and treated with enucleation.

Four years after the family's GGS diagnosis, the youngest daughter, aged 8, presented with similar complaints. Radiographic evaluation revealed large cystic lesions surrounding multiple impacted teeth in all four quadrants, reaching massive dimensions (Figure 1e, f). Genetic analysis confirmed a *PTCH1* gene mutation. Following marsupialization and enucleation, the lesions healed uneventfully. With orthodontic support, many permanent teeth associated with the cysts were preserved and brought into functional occlusion. No recurrence was observed during the 7-year follow-up period.

GGS is a rare autosomal dominant disorder primarily associated with *PTCH1* gene mutations.[1,2] Key clinical features include multiple jaw OKCs and early-onset BCCs.[3-5] Diagnosis is based on a combination of major and minor criteria, such as macrocephaly, skeletal anomalies, and congenital malformations.[4,6] Due to the high recurrence rate of OKCs, conservative surgical management requires long-term clinical and radiological follow-up.[2]

In this study, multiple OKCs and *PTCH1* mutations were identified in three related individuals, fulfilling established diagnostic criteria for GGS.[4,5] The detection of *PTCH1* mutations in all patients-despite reported mutation rates of 60%-85%-is noteworthy.[7] High penetrance of *PTCH1* mutations has been observed in familial GGS cases. Ramesh et al.[8] reported a detection rate of approximately 60%, with most mutations segregating within families. Another study found nearly 100% penetrance among offspring of mutation carriers. These findings support the high penetrance of *PTCH1* mutations in GGS and underscore the importance of early genetic screening.[9]

The recurrence observed in one case after 12 years highlights the need for prolonged follow-up in syndromic OKCs.

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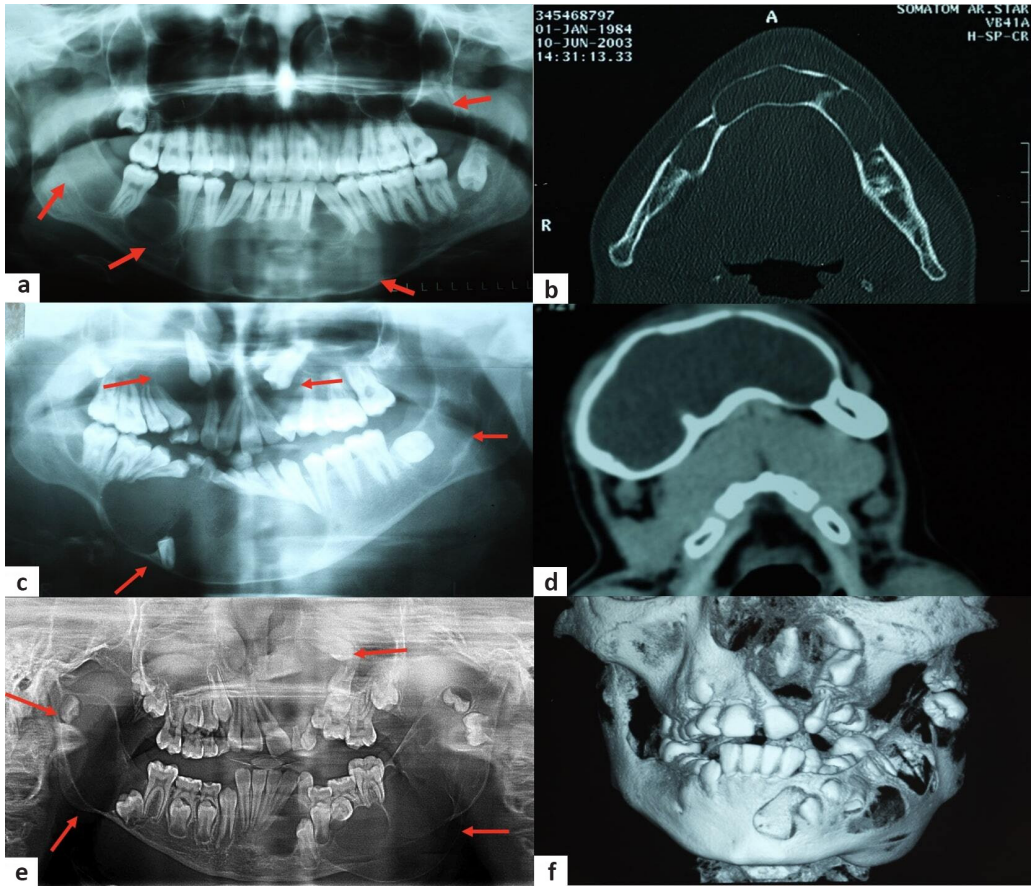

FIG. 1. (a) Patient 1, preoperative panoramic radiograph showing multiple radiolucent lesions extending from the right mandibular ramus to tooth 36 and another at tooth 28 (red arrows). (b) Axial CBCT image illustrating the mandibular lesion's extent. (c) Patient 2, preoperative panoramic radiograph showing radiolucent lesions in all quadrants with impacted teeth and jaw expansion (red arrows). (d) Axial CT showing buccolingual expansion due to a cystic lesion. (e) Patient 3, panoramic radiograph revealing lesions with impacted teeth in all quadrants. (f) 3D CBCT reconstruction displaying lesion extent and bone destruction.

CBCT, cone-beam computed tomography; CT, computed tomography; 3D, three dimensional.

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