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內文：

Introduction

1. Nevoid basal cell carcinoma syndrome (NBCCS), also known as Gorlin-Goltz syndrome.
2. Autosomal dominant disorder characterized by a predisposition to neoplasms and other developmental abnormalities.
3. Multiple basal cell carcinoma, keratocystic odontogenic tumors (KCOTs) in the jaws and bifid ribs.In addition to this triad, calcification of the falx cerebri, palmar and plantar epidermal pits, spine and rib anomalies, relative macrocephaly, facial milia, frontal bossing, ocular malformation, medulloblastomas, cleft lip and/or palate, and developmental malformations were also established as features of the syndrome.

Case report

1. 10 year-old white boy was first child of non-consanguineous parents of normal stature.
2. At the time of patient’ s birth, the father was 28 years old and the mother 25.
3. The syndrome did not affect the patient’ s parents and there were no familial antecedents.

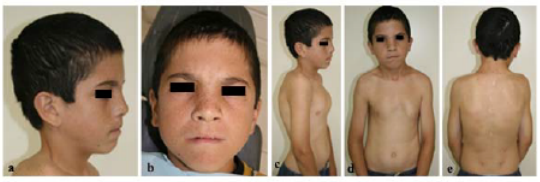

Figure 1 Clinical features of NBCCS. (a and b) Facial appearance of patient showed dysmorphic facial features, including relative macrocephaly (a) and ocular hypertelorism (b). (c and d) Lateral and frontal view showing pectum excavatum. (e) Vertebral anomaly characterized by cyphoscoliosis.

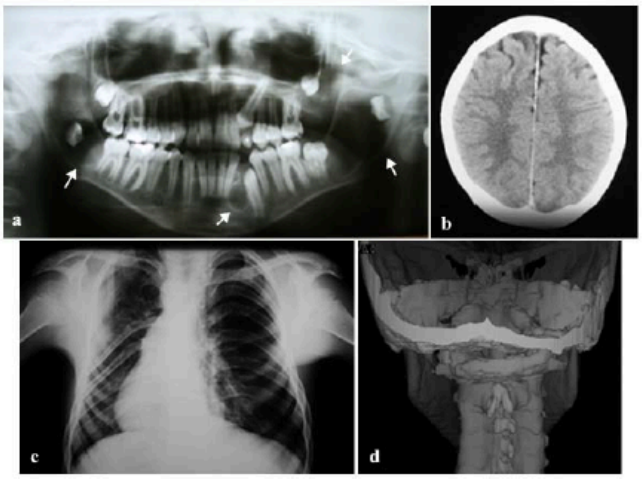

Figure 2 Imaging findings of NBCCS. (a) Orthopantographic examination suggesting the presence of multiple KCOTs in the maxilla and mandible (arrow). (b) Tomographic showing calcification of the cerebral falx. (c) Thorax film showing anomalies of the ribs characterized by flattening. (d) 3D tomographic reconstruction, showing spina bifida.

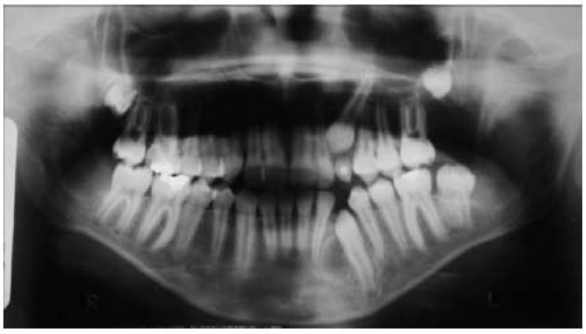

Figure 4 Orthopantographic examination profile at three months follow-up after the surgery to remove the cystic lesions.

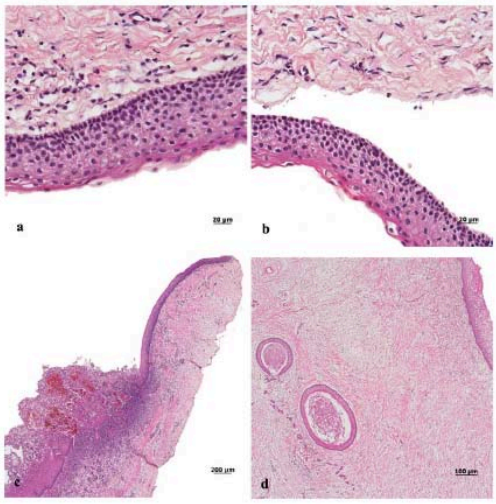

Figure 3 Histopathology findings of KCOT. (a and b) Prominent palisade basal cell layer with dark-staining nuclei and a corrugated surface with parakeratinization (H-E staining, original magnification × 40). (c) Presence of hyaline bodies and inflammatory changes that have destroyed parts of the lining ephitelium (H-E staining, original magnification × 4). (d) Prominent daughter cysts containing keratin whorls (H-E staining, original magnification × 10).

Discussion

1. Several studies have presented KCOTs, basal cell naevi and skeletal anomalies as the principal clinical features of NBCCS.
    a. Diagnostic criteria of NBCCS require the presence of two major, or one major and two minor criteria.
        i. Major criteria: presence of more than two basal cell carcinomas or one under the age of 20 years, histologically-proven KCOT of the jaw, cutaneous palmar or plantar pits, and bifid, fused or markedly splayed ribs.
        ii. Minor criterion: orofacial congenital malformations (cleft lip or palate, frontal bossing or moderate or severe hypertelorism), skeletal and radiological abnormalities (bridging of the sella turcica and vertebral anomalies), ovarian fibroma and medulloblastoma.
2. Development of multiple basal cell carcinomas, especially in the head and neck region.Not been possible to identify the presence of basal cell carcinomas.
3. NBCCS is caused by mutations in a tumor suppressor gene PTCH (human homologue of a Drosophila segment polarity gene Ptch) located in chromosome 9q22.
4. KCOTs are among the most consistent and common features of NBCCS. They are found in 65 to 100% of affected individuals.
5. The epithelial cells of the basal layer show increased mitotic activity, together with a potential for budding and the presence of daughter cysts in the wall. It has been reported that the presence of daughter cysts was related to the recurrence of KCOT. The mandible is involved more frequently than the maxilla and the posterior regions are the most commonly affected sites.
6. Treatment of KCOT, a conservative and an aggressive.
    a. In the conservative method, simple enucleation with or without curettage and marsupialization.
    b. Aggressive methods include peripheral ostectomy, chemical curettage with Carnoy’ s solution, and resection
7. In children who have yet to be erupted, conservative management should be

considered first because an aggressive operation can have an adverse effect on teeth development, the eruption process, and the development of the involved jaw.

8. Marsupialization followed by enucleation results in the lowest recurrence rate among the conservative treatment. Moreover, considering the complication of radical surgery, marsupialization followed by enucleation has been suggested as the conservative option for treatment of KCOT in younger patients.

9. Early diagnosis is important for counseling of patients to prevent harmful exposure to ultraviolet and ionizing radiations that increase the risk of developing basal cell carcinoma. The patient in this case study was sent to dermatologist for monitoring of possible skin lesions.

10. In order to be able to establish early diagnosis of NBCCS, specialists should carry out clinical and imaging examinations in early ages of life. Physicians and dentists must know the features of the syndrome well.

題號	**題目**
1	Where is gene location of PTCH that mutated cause NBCCS? **(A)** 9q22 **(B)** 10p22 **(C)** 9p21 **(D)** 10q22
答案(A)	**出處 :**Oral & Maxillofacial Pathology, Brad W. Neville, DDS, 2002
題號	**題目**
2	Which is the conservative treatment of Gorlin-Goltz syndrome? **(A)** simple enucleation **(B)** peripheral ostectomy **(C)** chemical curettage **(D)** resection
答案 (A)	**出處：**Oral & Maxillofacial Pathology, Brad W. Neville, DDS, 2002