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CASE REPORT

Nevoid Basal Cell Carcinoma (Gorlin) Syndrome (Case Report)

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Abstract

A16 year old boy was admitted to the hospital because of a swelling on the left side of mandible .He had multiple small indurated pearly papules on the head and neck with multiple well circumscribed radiolucent cystic space in mandible on panoramic radiography and CT-scan. Histopathologic studies of mandibular cyst and skin lesions revealed the finding in favor of nevoid basal cell carcinoma syndrome (NBCCS).

Keywords: Nevoid basal cell carcinoma; Gorlin syndrome; Keratocyst

Introduction

The autosomal dominant nevoid basal cell carcinoma syndrome (NBCCS), also referred to as Gorlin-Goltz Syndrome, represents a series of multiorgan abnormalities known to result from abnormalities in the PTCH gene. The approximate prevalence is reported to be 1 case per 56,000-164,000. The disease is present (inherited) at birth and most commonly manifests itself with either basal cell carcinomas (BCCs) (usually multiple) or Odntogenic Keratocysts (OKCs) presenting at the young age.[1,2]

Case Report

We report a 16 year old boy who came with a swelling on the left side of mandible 3 years ago. He didn't have any symptoms during mastication, and no history of dental infection or trauma. In clinical examination, irregular arrangements of teeth and disfigured jaw were seen. In dermatologic examination multiple small indurated pearly papules with telengiectatic vessels around, were present in the face and lateral aspect of the neck. One of them that became ulcerated and crusted was excised. In dental panoramic radiography, there was multiple well circumscribed radiolucency with smooth radiopaque margins in the mandible with unerupted dental component. (Figure 1) These findings were confirmed in CT scan. A grossly abnormal face with displacement of teeth in the mouth was seen. (Figure 2) When both maxillary sinuses were opened, it was found that these cysts contained pertinacious whitish material and multiple small teeth (Figure 3). Pathologic findings of the cysts, composed of uniformly thin epithelial lining, generally ranged from 8-10 layers thick. The basal layer exhibited a characteristic palisade pattern with polarized and intensely stained nuclei of uniform diameter. The luminal epithelial cells were parakeratinized and produced corrugated profile. The fibrous connective tissue component of the cyst wall was free of an inflammatory cell infiltration and relatively thin. The connection of epithelium and connective tissue was characteristically flattened with no epithelial ridge formation (Figure 4). The skin tumor lesion composed of nests and cords of cuboidal cells arose from the region of the epidermal basal cells. The neoplastic cells around the periphery of the invading nests were palisade. So the clinical manifestations of this case included multiple OKCs, facial bone defect and deformity and basal cell carcinoma. These signs are compatible with NBCCS.

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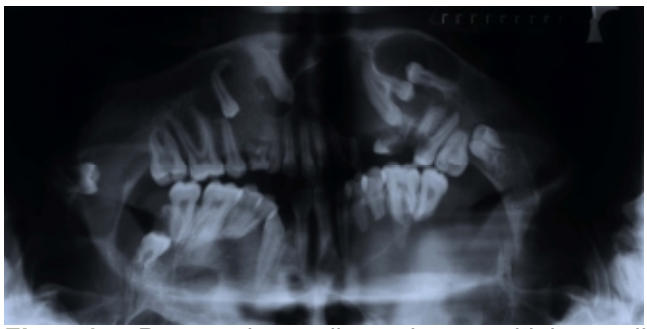

Fig. 1: Panoramic radiography, multiple well circumscribed radiolucency with unerupted dental component

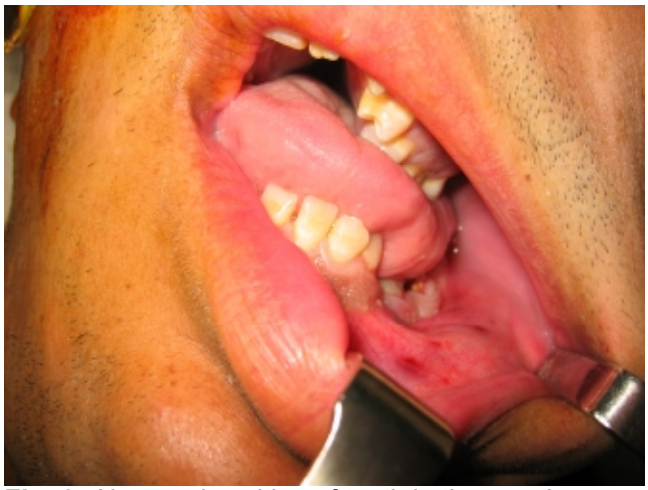

Fig. 2: Abnormal position of teeth in the mouth

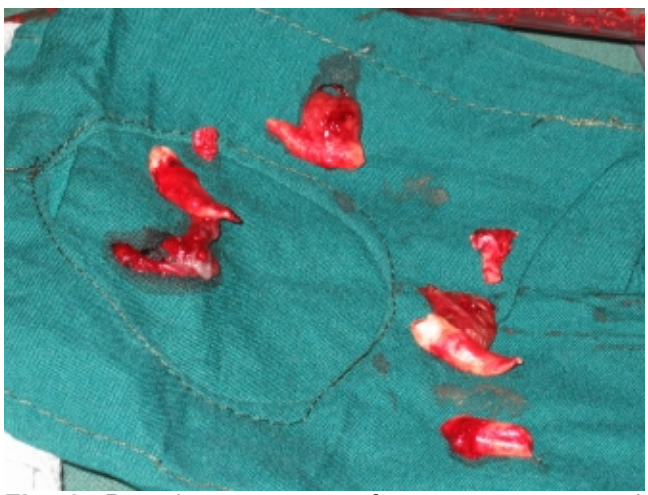

Fig. 3: Dental components of cysts were removed during surgery

Discussion

NBCCS is an inherited syndrome composed of multiple OKCs, bone defects and multiple BCCs. This syndrome is found in all races.[1,2] Men and women are affected equally. Despite the recent understanding of the underlying genetic basis of NBCCS, the diagnosis remains clinical. In several studies, Shanley[3] *et al.*, Evans[4] *et al.* and Kimonis[5] *et al.* have documented the incidence of the various features found in the syndrome. The disease has been reported by Pousti *et al.* presenting as recurrent mandibular cyst infection in Tehran, Iran.[6] Characteristic faces occur due to increased calvarial size.[4-7] Other contributing features include a broadened nasal root, frontal and biparietal bossing, mild hypertelorism, and an exaggerated length of the mandible. Ocular findings include congenital blindness due to corneal opacity and cataract or glaucoma, occurring in as many as 10-15% of patients.[4-7] BCCs are the most common finding in the syndrome; approximately, 97% of the patients have BCCs. These are most common on the face, the neck, and the upper part of the trunk, that is, in sun-exposed areas. Patients are particularly sensitive to ionizing radiation and ultraviolet (UV) light exposure, two important cofactor in developing BCCs.[5,7] The other cutaneous abnormalities include palmar and plantar keratotic pitting, multiple millia, and dermal calcinosis. Odontogenic keratocysts, which is a distinctive developmental odontogenic jaw cyst characterized by a parakeratinized stratified squamous epithelial lining, are seen in 74-80% of patients.[4-7] They usually begin to develop in the first decade. They are more common in the mandible than in the maxilla. Facial dysmorphogenesis involves a broad nasal bridge with ocular hypertelorism.[4-7]

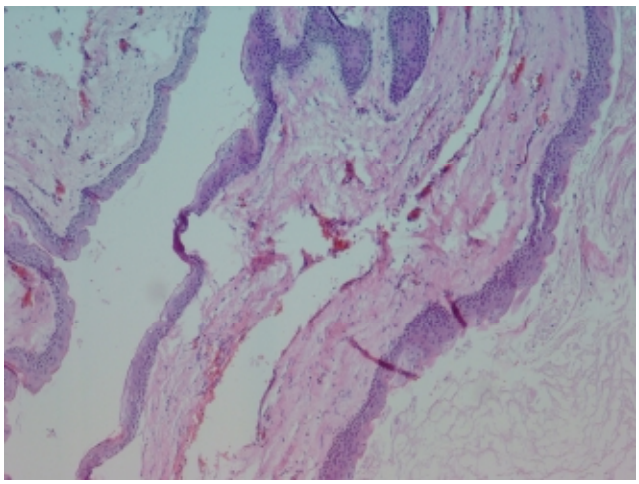

Fig. 4: Palisading of the basal cells and the epithelium maturing to a parakeratotic surface in keratocyst. (H&E × 400)

Neurologic abnormalities including meduloblastoma, dysgenesis and agenesis of the corpus callosum, calcinosis of the faix cerebri .Medulloblastoma (malignant tumors of the cerebellum) occur in 1-4% of the patients.[4-7] Skeletal abnormalities include polydactyly of the hands or the feet, hallux valgus may also more common. Calcification of the falx cerebri is the most common radiologic finding.[4-7] The defective gene associated with NBCCS is on chrososome 9p22.3, PTCH gene.[8,9] The protein product of the PTCH gene (a tumor suppressor gene) is a component of the hedgehog signaling pathway and is essential for development during embryogenesis and cell signaling in adults.[8,9] Mutation of PTCH gene is involved in the development of human syndromic basal cell carcinoma and is also present in the proportion of sporadic basal cell carcinoma and probably in some OKGs that occur sporadically.[8]

Surgical excision with peripheral osseous curettage is the preferred method of management. The recurrence rate of 10% to 30% of OKCs appears to be associated with several physical factors. The friable, thin connective tissue wall of the cyst may lead to incomplete removal.[1,2] Follow- up examinations is important for patients with this lesion. Patients should be evaluated for completeness of excision or new keratinocysts. In regard to skin cancer, patients should be advised to reduce UV light exposure and odontogenic keratocysts require regular dental follow-up visits.[5-7]

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Conflict of interest: None declared.

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