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Case Note

Multiple odontogenic keratocysts associated with Gorlin-Goltz syndrome

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Abstract

Gorlin-Goltz syndrome or Nevoid basal cell carcinoma syndrome is an autosomal dominant disorder with a predisposition to cancer. Features like basal cell carcinoma, odontogenic keratocysts, calcification of falx cerebri, bifid ribs, pits on palms and soles and hypertelorism are evident. A case of this rare disease seen on a 13 year old female patient is presented here, where multiple odontogenic keratocysts were causing disfigurement of the lower jaw as well as displacement and malocclusion of the lower teeth.

Key words: Nevoid basal cell carcinoma syndrome, Gorlin-Goltz syndrome, Odentogenic keratocyst, Calcification of falx cerebri.

Gorlin–Goltz syndrome which is also known as Nevoid Basal Cell Carcinoma Syndrome is a rare autosomal dominant disorder with strong penetrance and extremely variable expressivity. In 90% of the patients it is associated with multiple recurring odentogenic keratocyst(OKC)[1]. In addition to the jaw cysts, multiple nevoid basal cell carcinomas and skeletal anomalies are common findings[2]. The usual skeletal findings include bifid ribs, vertebral anomalies and frontal and temporo-parietal bossing. Other findings of this syndrome include hypertelorism, palmer and planter pits, calcification of falx cerebri, central nervous system and ocular lesions, cleft lip and palate, mandibular prognathism (class III jaw relationship) and in rare cases ovarian fibromas[3,4]. A case of Gorlin-Goltz syndrome is presented here in which most of the above mentioned findings are evident.

Case report

A 13 year old girl presented to the Dental Department at Kathmandu Medical College with complaints of swelling in the lower back gums and jaws since the last six months. On intraoral examination, her lower deciduous posterior teeth were still present and few permanent anterior teeth were missing. Because of the swelling, the buccal sulcus on both sides were obliterated and there was also expansion of the buccal cortical plates (Fig 1). The swelling had been increasing since the last 3-4 months and there was no pain associated with it, however the patient had parasthesia of the lower lip and chin.

An Orthopantomogram (OPG) was advised which revealed huge cyst like radiolucency with multiple loculi extending from the premolar region of one side of the jaw to the premolar region of the other (Fig 2). The erupting left third molar tooth also had a cyst like radiolucency over the crown extending up to the ramus of the mandible. Because of the pressure from the cyst, the permanent right lower canine and first premolar had been pushed towards the inferior border of the mandible and the left lateral incisor and canine were displaced (Fig 2). Due to the presence of multiple cysts like lesions in the jaw, Gorlin-Goltz syndrome was suspected and further investigations were carried out. A x-ray of the skull revealed calcification of falx cerebri (Fig 3) and a chest x-ray showed bifid ribs (Fig 4). Other findings included frontal bossing, hypertelorism (Fig 5a) and a Class III jaw relationship (Fig 1 and Fig b).

A FNAC was also carried out which was suggestive of odentogenic keratocyst. After discussing with the patients parents surgical removal of the cyst was planned.

Prior to the surgical procedure, an arch bar was placed on the lower jaw to prevent fracture of the mandible as

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well as to splint the unsupported permanent teeth. The cyst enucleation was done under general anesthesia via intraoral approach.

During the process eggshell crackling was evident which was suggestive of the thinned out cortical plates. After the cyst was enucleated, large areas of bone loss were seen and the displaced permanent teeth were visible on the floor of the cystic cavity (Fig 6). The enucleated cystic lining was sent for histopathological examination.

Carnoy's solution was then applied using cotton rolls for a period of five minutes. The excess solution was then irrigated using saline and the cavity was packed with Bismuth Iodine Paraffin Paste (BIPP).

The pack was removed after seven days and regular irrigation of the cavity was done every two days for the following two weeks. The wound healed uneventfully and the erupted teeth showed gradual improvement in their mobility.

OPG's were taken every three months and it was evident that bone regeneration was gradually taking place. Even the permanent teeth which were displaced downwards were erupting towards their respective normal positions in the lower jaw (Fig 7).

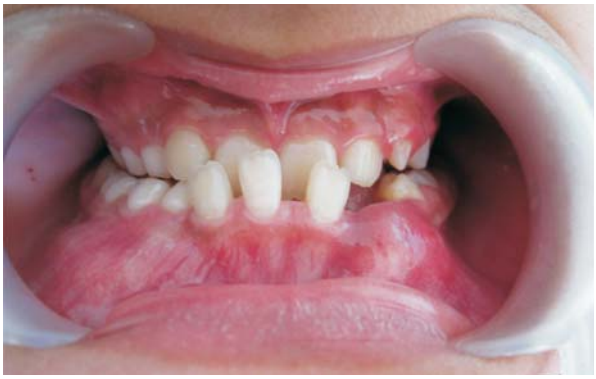

Fig 1: Swelling of the buccal cortical plates and displaced lower incisors

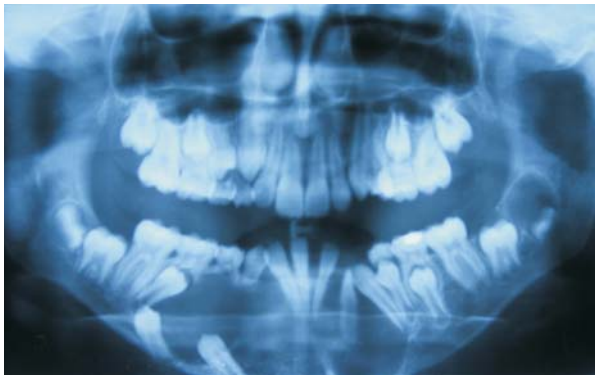

Fig 2: Preoperative OPG showing multiple cysts extending throughout the mandible and also the displaced lower permanent teeth

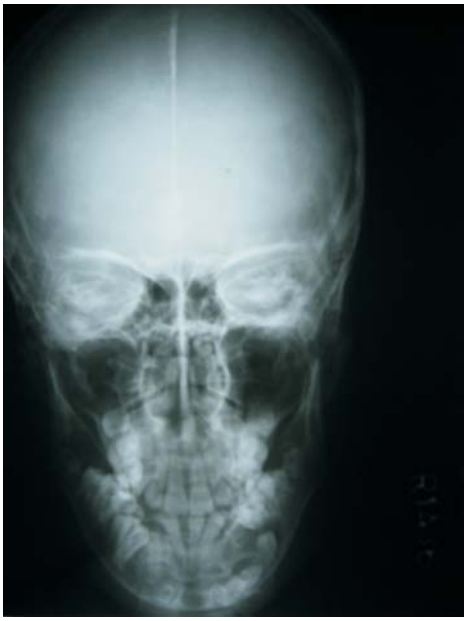

Fig 3: Calcification of the falx cerebri

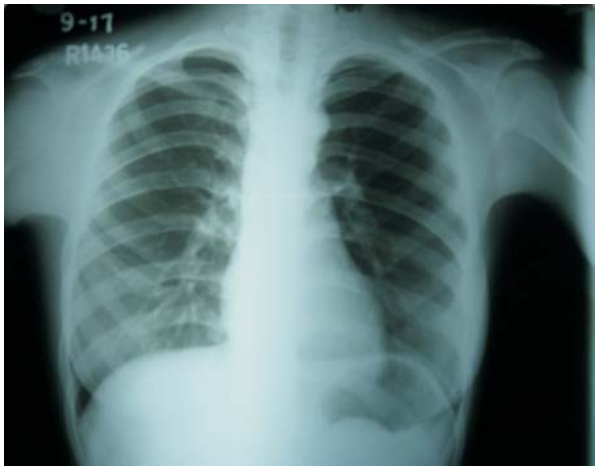

Fig 4: Chest x-ray showing bifid ribs

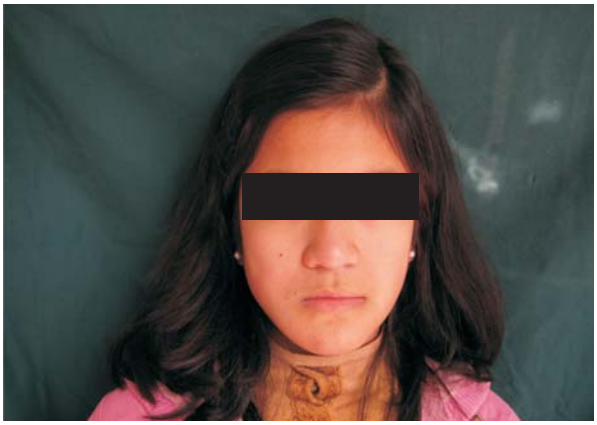

Fig 5a: Preoperative front profile photograph showing hypertelorism and frontal bossing

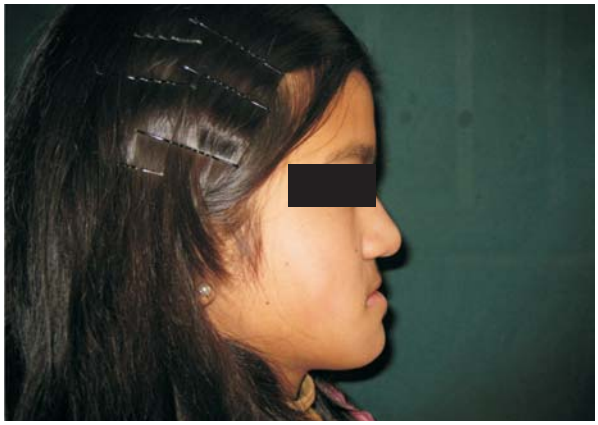

Fig 5b: Preoperative side profile photograph showing mandibular prognathism (Class III jaw relationship)

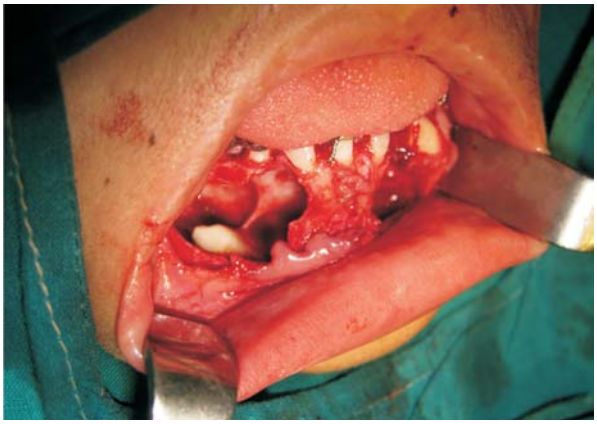

Fig 6: Extensive bone loss extending from one side of the jaw to the other seen after removal of the cyst. Very thinned out labial cortical plate and displaced right permanent canine are also seen.

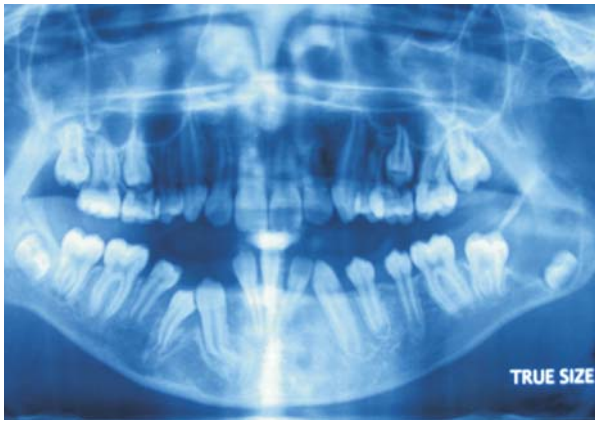

Fig 7: Post operative OPG (after 9 months) showing the erupting permanent teeth which were initially displaced and also the fully calcified bone in the region previously taken up by the cyst.

Discussion

The relationship of multiple dental cysts with other abnormalities was noted in the literature since the 1930's, but it was left to Gorlin and Goltz [4] in 1960 to tie all of this together into the syndrome that currently bears their name. In their article, they described the relationships of primodial cysts of the jaws with skeletal abnormalities including bifid ribs, hand and foot abnormalities and skin abnormalities including basal cell carcinomas, plamer pitting and various skin tags. Later it was recognised that the cysts found in the syndrome are the parakeratinised type of odentogenic keratocyst and that there are additional manifestations of the syndrome [5].

The prevalence of this condition is about 1 per 60,000 individuals and Gorlin-Goltz syndrome has been mapped to the long arm of chromosome 9 q22.3-q31[6,7,8]. Data also suggest that the product of this gene acts as a tumor suppressor and the syndrome's typical malformative pattern suggest that the main function is to control the growth and development of normal tissues. These abnormalities could determine malignant cutaneous tumors removing antineoplastic protection[9].

In the presented case, the histopathological report suggested OKC which is a common finding in Gorlin-Goltz syndrome. Histopathologically OKC is composed of parakeratinised epithelium, although rarely small foci of orthokeratinisation may also be found[10]. Parakeratinised areas show surface coagulation and there may be keratin in the lumen. There is a well defined basal layer composed of columnar or cuboidal cells[11]. The connective tissue wall of an OKC is composed of fibrous tissue rich in mucopolysaccharides and is free of inflammation[12].

Although Gorlin-Goltz syndrome is primarily recognised by multiple basal cell carcinomas, it was not evident in the above mentioned patient. The occurrence of basal cell carcinoma is in only 50% of the cases[13].

Removal of a large cyst, similar to the one seen in our case, occasionally weakens the remaining bony integrity and places it at a risk of pathologic fracture. This can be managed with intermaxillary fixation or placement of reconstruction plate. In our case, an arch bar was attached to the lower teeth to splint them as well as to support the remaining bone structure to prevent any fracture of the jaw.

Stoelinga, Voorsmit and colleagues, advocated excision of the overlying mucosa and have popularised the use of Carnoy's solution as a chemical tissue fixative[14,15,16,17].

The solution is a mixture of absolute alcohol, chloroform, glacial acetic acid and ferric chloride that penetrates the bone to a predictable, time dependant depth without injuring the neurovascular bundles. Application of the Cornoy's solution was done for about five minutes after the cystic lining was removed in our case. A five minute application penetrates bone to a depth of 1.54 mm, nerve to a depth of 0.15mm and mucosa to a depth of 0.51mm[15].

After enucleation of the cystic lining, the displaced permanent teeth were left untouched in the same position, because unerupted permanent teeth usually erupt towards the normal direction after removal of the obstruction in the erupting path. OPG's taken every 3 months showed that the displaced teeth were gradually becoming upright and were moving towards the alveolar ridge. After nine months from the surgical removal of the cyst the displaced right first premolar and canine and the left lateral incisor and canine have almost erupted to their normal respective positions (Figure 7). The patient was also relieved from the parasthesia of the lower lip and chin after removal of the cyst, because there was no more pressure on the mental nerve from the cyst.

Another cyst present above the crown of lower left third molar tooth also has to be removed and the patient and her parents have yet to make the decision for removal of the same.

Conclusion

Gorlin-Goltz syndrome is a condition which has a predisposition of cancer and it is very important to diagnose and treat the existing problems early. Thorough extraoral and intraoral examinations along with OPG, skull and chest radiographs help in proper diagnosis of the condition.

Odontogenic keratocysts of the jaws which can cause disfigurement of the face, mobility and even loss of teeth can be avoided by early detection and treatment of the problems.

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