

Delayed dental development in a patient with Gorlin syndrome: case report

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Gorlin syndrome is a relatively uncommon entity which was first reported by Ancell in 1841 in a patient with multiple benign cysts of the jaws.¹ In 1965, this condition took on its current name when Gorlin catalogued the related findings of 150 patients,² the most common of which were multiple nevoid basal cell carcinomas of the neck and face, rib and spine abnormalities, multiple cysts of the jaws, and lamellar calcifications of the falx cerebri.³ Since that time, the condition has been variously known as Gorlin-Goltz syndrome, Nevoid Basal Cell Carcinoma syndrome, and Basal Cell Nevus syndrome.⁴ This case presents an additional previously unreported finding of delayed dental development in a patient with Gorlin syndrome.

Literature review

Gorlin syndrome is inherited as an autosomal dominant trait with a high degree of penetrance and variable expressivity.⁵ The disorder has a population prevalence of 1 per 57,000 people, with a 3:1 male sex predilection.⁶ There are more than 40 different diagnostic findings associated with Gorlin syndrome. The best known feature is the presence of multiple basal cell carcinomas, which typically appear between puberty and 35 years of age,⁷ covering the trunk, neck, cheeks, nose, and eyelids of affected individuals.³ The number of these lesions can vary from a few to several thousand, varying in size from 1 to 10 mm in diameter.⁷

The second most common diagnostic finding in Gorlin syndrome is the presence of multiple odontogenic keratocysts (OKCs), which occur in up to 75% of all cases. These cysts have a peak occurrence in the second and third decade of life and are more common in the body and ramus of the mandible (80%) than in the maxilla. The OKCs may vary in size, be found singularly or in groups, and be unilateral or bilateral.⁵ In young patients, the OKCs can cause displacement of developing permanent teeth, expansion of the bone, be associated with an unerupted tooth, and occasionally cause root resorption.⁶

Radiographically, the OKC appears as a well-circumscribed radiolucency with smooth or scalloped

margins and a thin opaque border. They may be either unilocular or multilocular in appearance.⁵

The OKCs have been reported to have a recurrence rate of between 20 and 60%, with a peak recurrence rate within the first 5 postoperative years.⁸ Treatment of odontogenic keratocysts usually includes complete enucleation with aggressive curettage to ensure removal of accessory daughter cysts that can be found in and adjacent to the walls of the parent cyst.⁹

The skeletal components of patients with Gorlin syndrome include a characteristic marfanoid appearance as well as frontoparietal bossing, a low occiput, and mild hypertelorism. More than 85% of patient's with Gorlin syndrome exhibit lamellar calcification of the falx cerebri. Additional skeletal defects may include anteriorly splayed, fused, partially missing, hypoplastic, or bifid ribs.¹⁰

The neurologic anomalies associated with Gorlin syndrome are agenesis of the corpus callosum, congenital hydrocephalus, bridged sella turcica, medulloblastoma, seizure activity, and mental retardation.¹⁰

The dental components of Gorlin syndrome include the odontogenic keratocysts of the jaws, mandibular prognathism, and/or cleft lip and palate.⁶ While many of these findings will be found in conjunction with delayed dental development, to date there have been no reports of patients with Gorlin syndrome who have significantly delayed dental exfoliation, eruption, and development as a component.

Case report

A 14-year, 9-month-old Caucasian male presented to the University of Maryland Pediatric Dentistry Emergency Clinic complaining of increasing difficulty with chewing. A review of the patient's past medical history was unremarkable.

On physical examination, the patient was found to have an asymptomatic expansile swelling of unknown duration in the left anterior maxilla (Figs 1, 2), which was causing an extraoral obliteration of the patient's left nasal cheek fold. No signs of trigeminal or facial parasthesia were observed.

The patient exhibited significantly delayed dental ex-

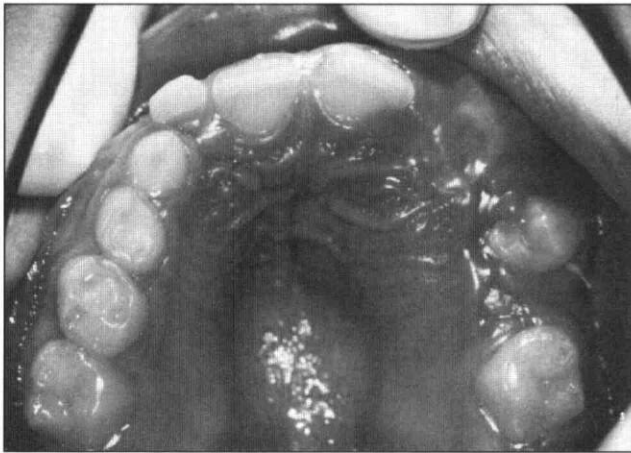


Figure 1: Preoperative clinical photograph of maxillary anterior cyst—occlusal view.

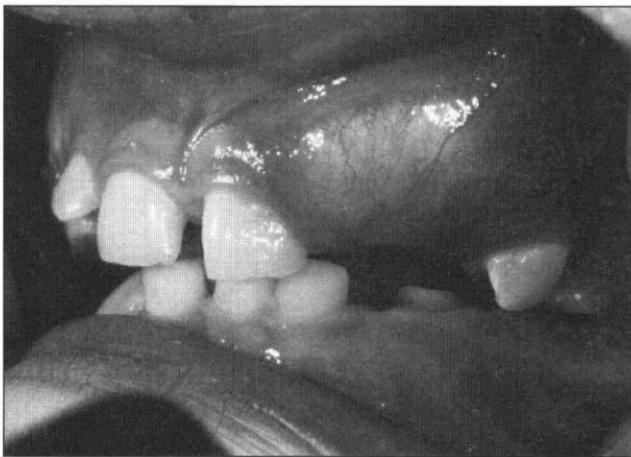


Figure 2: Preoperative clinical photograph of maxillary anterior cyst—lateral view.

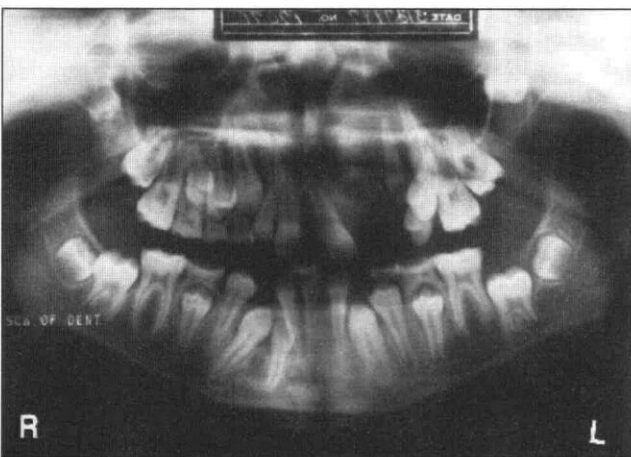


Figure 3: Preoperative panorex radiograph showing maxillary anterior radiolucency, maxillary left and right posterior expansion between the second and third permanent molars, delayed dental development and dilacerated root on the mandibular right lateral incisor.

foliation, eruption, and development for a 14-year, 9-month-old Caucasian male. The maxillary right first and second primary molars, mandibular left first and second primary molars, and mandibular right second primary molar were all over-retained and nonmobile. There was no clinical evidence of the permanent left maxillary lateral incisor, cuspid, or second premolar. The patient's maxillary left permanent first premolar was found to be partially erupted and exhibited Class II mobility. The patient's permanent second and third molars were not clinically visible, nor was there any evidence of the patient's mandibular right lateral incisor or cuspid.

Additionally, on physical examination, the patient was found to have several palmar and plantar pits as well as mild frontoparietal bossing. The patient's mother exhibited multiple basal cell carcinomas about the head and neck, although none were found on the patient.

On radiographic examination Fig 3, a 3.0 x 2.5-cm radiolucency with poorly defined borders was seen in the left anterior maxilla, which was causing significant displacement of the adjacent permanent left lateral incisor and cuspid, without evidence of obvious root resorption. In addition, there was superior and lateral displacement of the floor of the maxillary sinus in the anterior region. The posterior wall of the maxillary anterior cyst was shown to have displaced the root of the maxillary left first premolar distally and laterally over the developing second premolar, preventing the second premolar from erupting.

The Panorex[®] also revealed a bilateral expansion between the impacted maxillary second and third molars, with a poorly defined radiolucency surrounding the maxillary left third molar. In addition, there appeared to be a pericoronal radiolucency associated with the unerupted mandibular permanent right canine. The mandibular permanent right lateral incisor was impacted and found to have a dilacerated root, which was preventing its eruption.

Based on these previously undiagnosed findings, the patient was referred to the Oral-Maxillofacial Surgery service at the University of Maryland Hospital. As part of the patient's surgical work-up a head and neck CT scan was ordered, which revealed three separate, well-defined cystic areas of bone expansion in the left posterior, left anterior, and right posterior maxilla (Fig 4).

The patient's lateral skull film showed a calcified falx cerebri and his chest film revealed evidence of a bifurcation in the right seventh rib. Vertebral examination revealed no spinal deformities or abnormalities.

In addition to the patient's generalized delayed eruption of permanent teeth and delayed exfoliation of primary teeth, the unerupted permanent teeth appear fully developed on radiographs without any subsequent

primary tooth root resorption. A differential diagnosis for these dental findings may include systemic diseases such as hypothyroidism, hypoparathyroidism, hypopituitarism, or Albright's hereditary osteodystrophy (pseudohypoparathyroidism). The patient's baseline laboratory studies were all within normal limits; however, the patient needs to be referred for a more thorough endocrine work-up before these systemic diseases can be eliminated as possible etiologies for the dental presentation.

The patient's left maxillary anterior lesion was subsequently biopsied and sent for histologic evaluation. A differential diagnosis was developed including primordial cyst, dentigerous cyst, ameloblastoma, adenomatoid odontogenic tumor (AOT), odontogenic myxoma, calcified odontogenic cyst, and calcifying epithelial odontogenic (Pindborg) tumor as well as OKC.⁹

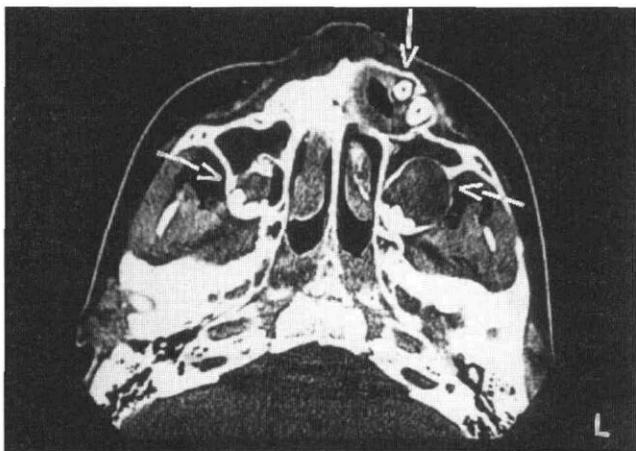


Figure 4: Computed tomography (CT) image revealing three separate, well-defined areas of bone expansion in the left posterior, left anterior, and right posterior maxilla. The left anterior cyst contains the left lateral incisor and canine.



Figure 5: Maxillary anterior cyst with accessory daughter cysts after removal.

The results of the biopsy confirmed that the specimen was an odontogenic keratocyst.

This patient's history and physical examination revealed three major (multiple OKCs, palmar/plantar pits, and calcified falx cerebri) as well as two minor (bifid rib and frontoparietal bossing) diagnostic criteria to satisfy the diagnosis of Gorlin syndrome.¹¹

The patient was admitted to the hospital for total cyst enucleation with aggressive curettage which was performed under general anesthesia. The surgical procedure included removal of the patient's maxillary left and right posterior cysts along with the impacted second and third molars associated with them. The maxillary left anterior cyst was removed along with the lateral incisor and cuspid that were contained within the cyst cavity. Several accessory daughter cysts were removed adjacent to the wall of the maxillary anterior parent cyst Fig 5.

The five over-retained primary molars were removed, as well as the dilacerated mandibular lateral incisor. The pericoronal radiolucency associated with the unerupted mandibular permanent right canine was believed to be a hyperplastic dental follicle, not an early OKC, and the decision was made to follow it on recall. The patient was discharged the following day having tolerated all procedures well without complications.

The patient was seen in the Pediatric Dental Clinic at the University of Maryland Dental School for an evaluation 1 week postoperatively and found to be healing well. There was significant mobility associated with the maxillary left first premolar, however, splint therapy was not recommended due to the patient's postoperative level of discomfort as well as the long span needed between abutment teeth in order to achieve proper immobilization. The patient was given home care instructions and was to be seen in the clinic 3 months postoperatively for prosthetic and orthodontic evaluations.

The patient did not present to the clinic for his 3-month recall appointment and all attempts to locate and contact the patient have been unsuccessful. At this point, the patient is currently lost to any follow-up.

Discussion

Gorlin syndrome is a rare but significant entity that affects the cranial, maxillofacial, cutaneous, and musculoskeletal systems of the body. The management of patients with Gorlin syndrome depends on the specific defects, anomalies, and pathologic findings which are manifested and to what extent they are expressed.

The dental findings of patients with Gorlin syndrome include odontogenic keratocysts, mandibular prognathism, and/or cleft lip and palate.⁴ While many of these dental conditions may be associated with delayed dental and oral development, there has not pre-

viously been a report of a patient with Gorlin syndrome with delayed primary tooth exfoliation and permanent tooth eruption as a significant dental finding.

This patient's dental findings may be a manifestation of a systemic disease, and not at all related to Gorlin syndrome. Additional endocrine studies are necessary in order to completely eliminate the possibility of hypothyroidism, hypoparathyroidism, hypopituitarism, or Albright's hereditary osteodystrophy as possible etiologies. Additionally, the patient's unusual dental presentation may merely be a coincidental finding, and not at all associated with the presence of Gorlin syndrome. More patients with Gorlin syndrome will have to be evaluated by pediatric dentists and endocrinologists in order to confirm this finding.

While this patient has currently been lost to follow-up, it is anticipated that the dentition will continue to develop. The maxillary left first and second premolars, which were found outside the wall of the anterior cyst, were retained in the hope that the cavity would fill in with bone. The first premolar would then continue to erupt and could be orthodontically repositioned. This would allow for the normal eruption of the patient's maxillary second premolar and a more stable arch form with less severe prosthetic consequences. Eventually, the patient's posterior occlusion and anterior segment can be properly restored using an implant retained prosthesis or a removable partial denture. As this patient is currently lost to follow-up, I can't state what dental events actually occurred.

A patient with Gorlin syndrome should be monitored annually by a dental and medical team to evaluate for potential cyst recurrences, new cyst formations, and/or the development of basal cell carcinomas that may require additional treatment.¹²

Because of the multisystem involvement and variable expressivity of Gorlin syndrome, these patients must be evaluated by many medical and dental specialists in order to properly sequence their treatment. As many of these patients will be seen and subsequently

diagnosed by pediatric dentists, a thorough understanding of the various components of the syndrome is essential. Delayed dental development in a patient with Gorlin syndrome is a new finding that requires attention and appropriate management in order to provide the patient with comprehensive treatment.

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