ARTICLE

Triona McNamara, BDS, FDS, DOrth, Carroll-Ann Trotman, BDS, MA, MS, Kathleen A. Russell, DDS, MSc

Gorlin-Goltz: What's in a name?

This paper describes the clinical features of two very distinct syndromes with similar names: Gorlin-Goltz and Goltz-Gorlin Syndromes. A case report is presented that highlights the differences between these syndromes. To avoid errors in diagnosis because of the similarity in names, the authors caution that, based on additional information now available, the preferred names should be Focal Dermal Hypoplasia syndrome for Goltz-Gorlin syndrome and Nevoid Basal Cell Carcinoma syndrome for Gorlin-Goltz syndrome.

The syndromes Gorlin-Goltz and Goltz-Gorlin are very distinct; each syndrome has very specific oral findings. The similarity in nomenclature, however, has led to some confusion in the literature. As an example of the confusion that exists in the literature, Witschel[1] recently reported a case by Manzi[2] that had confused these two syndromes. Witschel described a patient with oral manifestations of Gorlin-Goltz Syndrome, and demonstrated the difficulties and differences in dental management of the Gorlin-Goltz syndrome relative to the Goltz-Gorlin Syndrome.

Gorlin-Goltz Syndrome, or Nevoid Basal Cell Carcinoma Syndrome (NBCCS), was described initially by Jarisch in 1894.[3] In 1960, Gorlin and Goltz[4] classified NBCCS as a distinct syndrome with three main characteristics: multiple basal cell carcinomata, odontogenic keratocysts, and rib anomalies. Subsequently, other associated anomalies were found. These anomalies involved the neurological system, with calcification of the falx cerebri and tentorium cerebelli, bridging of the sella turcica, mental retardation, medullablastoma, and hydrocephalus; the cardiac system, with cardiac fibromas and valvular defects that were reported in up to 15% of cases; the ocular system, with hypertelorism, strabismus, ptosis, congenital cataracts, and glaucoma; and the skeletal system, with widespread involvement of the ribs, vertebra, and digits, an increased stature, and a Marfanoid build. In addition, ovarian fibrosarcoma and hypogonadism have been described in females and males, respectively.

Inheritance is believed to be autosomal-dominant with high penetrance and variable expressivity.[5-8] In NBCCS, the nevoid basal cell carcinomas generally appear between puberty and 35 years of age and have the following features: They do not tend to be aggressive before puberty. They may be few or number in the thousands; and they may arise on the skin—especially of the face, neck, and upper trunk. But they are rare on the extremities, abdomen, and lower trunk. In addition, multiple odontogenic keratocysts develop during the first decade of life, unlike isolated odontogenic keratocysts that develop later. Usually, these multiple keratocysts are asymptomatic and are found most commonly in the mandibular third molar and maxillary canine regions.[5-9]

Case report

A 12-year-old boy was referred to our clinic for treatment of a dental abscess. The patient's past medical history included arrested hydrocephalus, mild mental retardation, epilepsy, and scoliosis. A number of basal cell nevi were present in the patient's upper trunk. In addition, at ten years of age, the patient had surgery for the removal of a basal cell nevus because of concerns regarding its changed nature. The patient's past dental history was one of poor compliance with routine dental care. He had had previous oral surgery at another center to enucleate an odon-

togenic keratocyst in the left mandibular premolar region, but had no prior history of any dental extractions. A comprehensive family history could not be ascertained, but his brother was also affected with this condition.

On extra-oral clinical examination, the patient had a skeletal and dental Class III malocclusion with a prognathic mandible. Intra-orally, an abscess was found associated with a partially erupted maxillary right permanent canine. There was a distal deflection of the canine with expansion of the associated buccal bone, and pus exuding from the region (Fig 1). In addition, there were delayed dental development, hypodontia, and several caries lesions.

On radiological examination, the right mandibular second premolar, right maxillary first and second premolars, left maxillary second premolar, and right maxillary third molar were seen to be missing. There were well-defined radiolucent lesions and associated bony expansions in the maxillary right canine region, mandibular incisor and canine regions, and in the mandibular left third molar region, consistent with odontogenic keratocysts (Figs 2, 3). Also, there were disruptions in the eruption patterns of teeth adjacent to cystic lesions; the mandibular left canine and left third molar were affected, as well as the maxillary right canine. The maxillary left permanent canine and first premolar were transposed, with delayed dental development of the maxillary first premolar.

Treatment at the time of presentation involved surgical removal of the offending tooth, the maxillary right permanent canine, and the associated cyst. The results of a biopsy report on the cystic tissue confirmed the presence of an infected odontogenic keratocyst associated with the maxillary right permanent canine. The continued management of this patient's condition involved additional surgeries to treat both the maxillary and mandibular cysts, and the treatment of dental caries. He continues to be monitored carefully, but due to his ongoing oral surgery needs and inability to comply easily with dental procedures, orthodontic treatment to correct his malocclusion has not been considered.

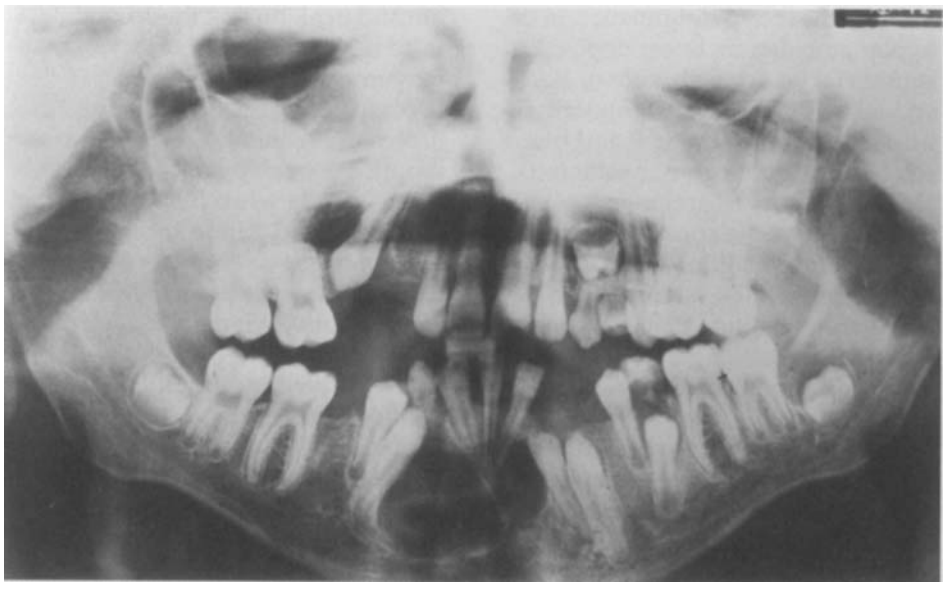

Figure 1. Intra-oral view showing disruption of the eruption of the maxillary right permanent canine buccal bony expansion associated with an infected odontogenic keratocyst, and incisor Class III relationship.

Discussion

Goltz Syndrome, or Focal Dermal Hypoplasia (FDH), was described first as a definitive syndrome by Goltz-Gorlin in 1962,[11] with the earliest case report by Jessner in 1921.[12] FDH is characterized by widespread anomalies of both ectodermal and mesodermal structures that involve the skin, eyes, mouth, central nervous system, and musculoskeletal system. Unlike NBCCS, the signs and symptoms of FDS are apparent at birth and may be life-threatening. Like NBCCS, the cause of FDH is unknown;

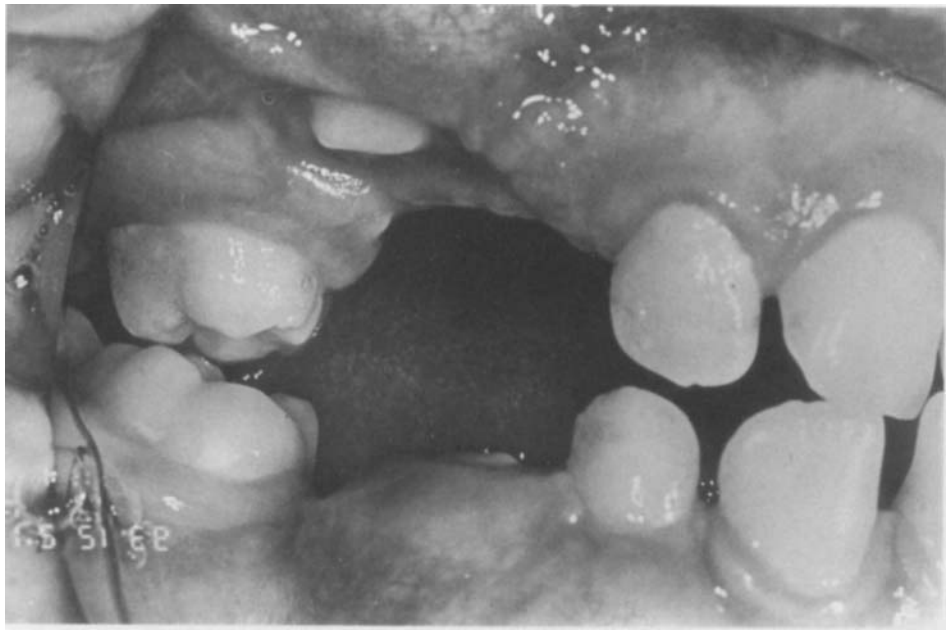

Figure 2. Orthopantomograph (radiographic view) showing hypodontia, delayed loss of primary teeth, transposition, and odontogenic keratocysts in the mandibular incisor and left third molar regions.

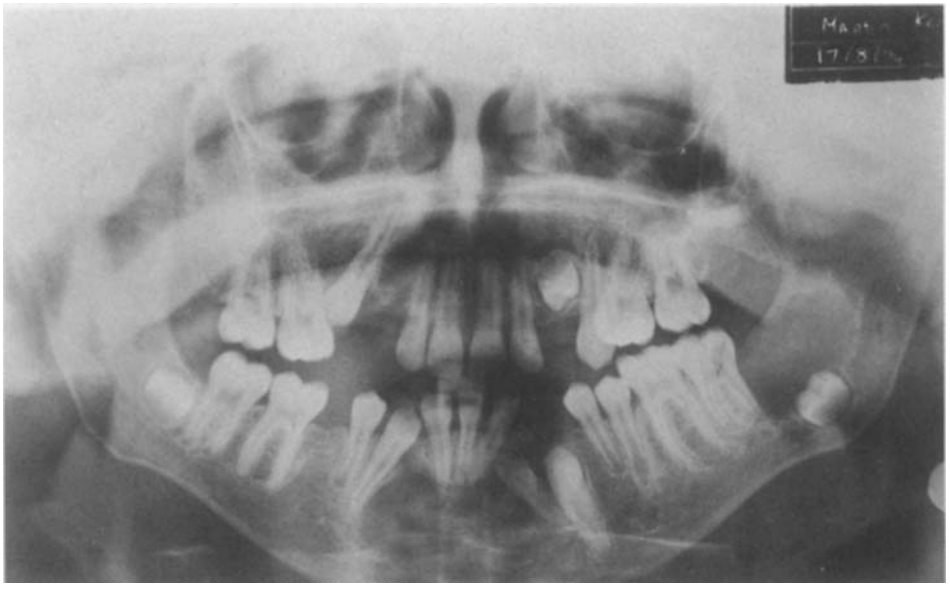

Figure 3. Orthopantomograph (radiographic view) taken approximately eight months later, showing significant enlargement of the odontogenic keratocyst in the mandibular left third molar region with gross displacement of this tooth.

approximately 95% of reported cases are sporadic, and only 10% affect males. In those cases that are associated with a familial history, it has been suggested that the disorder is X-linked-dominant, and usually, the transmitting parent is affected mildly. FDH tends to be lethal in males; in females, fertility may be reduced significantly. Skin defects have been reported in FDH that are very different from those of NBCCS, and include linear hyperpigmentation or hypopigmentation, linear deposits of superficial fat, focal absence of skin, and telangiectasis. Ocular abnormalities—such as chorioretinal and iris colobomata, strabismus, nystagmus, blocked tear ducts, and microophthalmia—have also been described. Skeletal abnormalities associated with the extremities and scoliosis may occur, resulting in a short and asymmetric stature.[13-15]

The oral abnormalities in FDH have been well-documented and are very distinct from those of NBCCS. Widespread papillomas of the oral mucosa have been reported that may extend to the esophageal region. Cleft lip and palate have also been described, and dental anomalies such as hypodontia, oligodontia, enamel pitting, delayed eruption, and orthodontic malocclusion have been reported.[15,16] Dental abnormalities of FDH are primarily poor tooth structure that may be further complicated by hypodontia. Poor crown and root formation renders the teeth unesthetic and of uncertain prognosis. Optimal dental care involves complex and comprehensive restorative treatment.

NBCCS' significant oral findings are odontogenic keratocysts that generally develop in the first decade of life and peak during the second or third decade. These keratocysts may remain symptom-free but can become very large before being detected. Frequently, as in this case report, the odontogenic keratocyst is detected only when it becomes secondarily infected. These keratocysts can disrupt the development of adjacent teeth. In this case report, over an eight-month period, the development of the mandibular left third molar was disrupted by a rapidly forming keratocyst (Figs 2, 3).

In summary, Gorlin-Goltz and Goltz-Gorlin (Goltz Syndrome, as it is known commonly) are two independent syndromes. Both have abnormalities affecting tissues of similar origin, and demonstrate significant oral and dental findings. As reported in this paper, errors can be made in diagnosis because of the similarity in the names. Gorlin, in 1971,[10] advised against using eponyms. With additional information now available, Gorlin suggests that the preferred names be Focal Dermal Hypoplasia Syndrome for Goltz-Gorlin Syndrome, and Nevoid Basal Cell Carcinoma Syndrome (NBCCS) for Gorlin-Goltz Syndrome. NBCCS is suggested despite the fact that not all affected adults with NBCCS have basal cell carcinomas.[13] "To name" is defined as "to assign, to state, give particulars of, to specify".[17] A name is also defined as "a word or term by which a person or thing is known".[18] Should Gorlin-Goltz Syndrome and Goltz Syndrome be the names used to decribe these syndromes? Care is advised.

Dr. McNamara is a Consultant Orthodontist, Regional Orthodontic Department, Eastern Health Board, St James's Hospital, Dublin 8, Ireland. Dr. Trotman is an Assistant Professor in the Department of Orthodontics and Pediatric Dentistry, University of Michigan, Ann Arbor, Michigan 48109-1078. Dr. Russell is an Assistant Professor in the Department of Orthodontics, Dalhousie University, Halifax, Nova Scotia, Canada.

1. Witschel H. Letter to the editor. Ophthalmologica 202:115, 1991.
2. Manzi G, Magli A, Pignalosa B, Liguori G. The Gorlin-Goltz syndrome: case report. Ophthalmologica 200:104-6, 1990.
3. Jarisch W. Zur Lehre von der Hautgeschwulsten. Arch Dermatol Syph (Berlin) 28:162-222, 1894.
4. Gorlin RJ, Goltz RW. Multiple nevoid basal cell epithelioma, jaw cysts and bifid rib syndromes. N Engl J Med 262:908-12, 1960.
5. Gorlin RJ. Nevoid basal-cell carcinoma syndrome. Medicine 66:96-113, 1987.
6. Sun DX, Yu Q, Wang PS, Qian GQ. Basal cell carcinoma syndrome: report of 10 cases. Dentomaxillofac Radiol 19:181-4, 1990.
7. Lovin JD, Talarico CL, Wegert SL, Gaynor LF, Sutley SS. Gorlin's syndrome with associated odontogenic cysts. Pediatr Radiol 21:584-7, 1991.
8. Posnick JC, Clokie CML, Goldstein JA. Maxillofacial consideration for diagnosis and treatment in Gorlin's syndrome: access osteotomies for cyst removal and orthognathic surgery. Ann Plast Surg 32:512-8, 1994.
9. Evans DGR, Ladusans EJ, Rimmer S, Burnell LD, Thakker N, Farndon PA. Complications of the naevoid basal cell carcinoma syndrome: results of a population based study. J Med Genet 30:460-4,

1993.
10. Gorlin RJ, Heddie SO. The multiple nevoid basal cell carcinoma syndrome revisited. Birth Defects: Orig Art Ser 7:140-8, 1971.
11. Goltz RW, Peterson WC, Gorlin RJ, Ravits HG. Focal dermal hypoplasia. Arch Dermatol 86:708-17, 1962.
12. Jessner M. Falldemonstration Breslauer dermatologische Vereinigung. Arch Dermatol Syph (Berlin) 133:48, 1921.
13. Gorlin RJ, Cohen MM, Levin LS. Syndromes of the head and neck. 3rd ed. Oxford: Oxford University Press, pp. 472-5, 1990.
14. Goltz RW, Henderson RR, Hitch JM, Ott JE. Focal dermal hypoplasia syndrome. Arch Dermatol 101:1-11, 1970.
15. Goltz RW. Focal dermal hypoplasia syndrome: an update. Arch Dermatol 128:1108-11, 1992.
16. Mallory SB, Krafchik BR. Goltz syndrome. J Pediatr Dermatol 6:251-3, 1989.
17. The Oxford English Dictionary. 2nd ed. Oxford: Clarendon Press, pp. 204-5, 1989.
18. Collins New Compact English Dictionary. Harper Collins Publishers, p. 555, 1992.