Korean Journal of Oral and Maxillofacial Radiology 2004; 34 : 203-7

Recurrent odontogenic keratocysts in basal cell nevus syndrome: Report of a case

Byung-Do Lee, Jin-Hoa Kim, Dong-Hoon Choi, Kwang-Soo Koh, Sang-Rae Lee*

Department of Oral and Maxillofacial Radiology, School of Dentistry, Wonkwang University
**Department of Oral and Maxillofacial Radiology, School of Dentistry, Kyunghee University*

ABSTRACT

Basal cell nevus syndrome (BCNS) is principally characterized by cutaneous basal cell carcinomas, multiple odontogenic keratocysts and skeletal abnormalities. Our patient represented several characteristics of BCNS, such as, multiple odontogenic keratocysts, facial nevus, calcification of falx cerebri, parietal bossing and mental retardation. The cyst on posterior mandible showed recurrent and newly developing tendency. ***(Korean J Oral Maxillofac Radiol 2004; 34 : 203-7)***

KEY WORDS : Odontogenic Cysts; Nevus; Carcinoma

Basal cell nevus syndrome (BCNS) is a hereditary condition transmitted by autosomal dominant trait.[1, 2] Jarish[3] first described many symptoms, Howell and Carlo[4] associated basal cell nevus with cutaneous cancers and various anomalies. Gorlin and Goltz[5] defined this syndrome comprising the principal triad of multiple basal cell nevi, multiple odontogenic keratocyst (OKC), and skeletal abnormalities. This disorder has been variously known as basal cell nevus syndrome, nevoid basal cell carcinoma syndrome, Gorlin-Goltz syndrome.[6]

Because BCNS have varying expressions, the criteria for diagnosis are not clear and it seems that the syndrome may occur in a minor form without either basal cell carcinomas or OKC.[7]

Multiple OKC are well-recognized feature of BCNS,[8] and there have been many reports concerning recurrence of OKC, that was the new and recurring cysts at increasing frequency until about age 30, when the rate of development tends to decrease.[9-11]

Our patient showed several characteristics of BCNS and recurrent tendency of OKC.

Case report

19-Year old male visited Wonkwang dental hospital at 2001-05-21 for the chief complaints of left submandibular swelling. The C.C onset was about 7 days ago. There were 4 unilocular radiolucent lesions on panoramic view (Fig. 1), those were on mandibular anterior region, right and left mandibular posterior area, and left maxillary posterior area. These lesions showed well defined border, especially cortical expansion and thinning on lower anterior lesion (Fig. 2). Dome shaped radiopacity with well defined border of left maxiilary sinus on Waters' view suggesting cystic lesion (Fig. 3). From these radiographic finding, we consider multiple OKC and the possibility of BCNS.

Intracranial calcifications of the falx cerebri and parietal bossing were evident on skull PA view (Fig. 4). We observed compound nevus on his face and abnormality of left thumb. Additionally he showed mental retardation but we cannot confirm familial history.

The jaw cystic lesions were marsupialized for several months for size reduction.

At 2002-02-25, enucleation of cystic lesions, iliac bone graft procedure was done on lower anterior area. Fig. 5 showed reduced radiolucency of pre-cystic lesions after operation. But we found newly developed and recurrent cystic lesions at right and left mandibular posterior area on radiograms of 2004-05-06 (Figs. 6, 7). Biopsy was done and showed the findings of odontogenic keratocyst (Fig. 8).

From these clinical, radiographic, and histopathological finding, this patient was diagnosed as basal cell nevus syndrome with recurrent OKC.

Discussion

BCNS is a rare disease. The prevalence has been estimated about 1 per 60, 000.[12] This disorder has an autosomal dominant mode of inheritance.[2,13] Gene on chromosome 9q (22.3-

Received October 6, 2004; accepted November 1, 2004
Correspondence to : Prof. Byeong-Do Lee
Dept of Oral and Maxillofacial Radiology, College of Dentistry, Wonkwang University, 344-2, Shinyong dong, Iksan city, Chunbuk, 570-711 Korea.
Tel) 82-63-850-1912, Fax) 82-63-857-4002, E-mail) eebydo@wonkwang.ac.kr

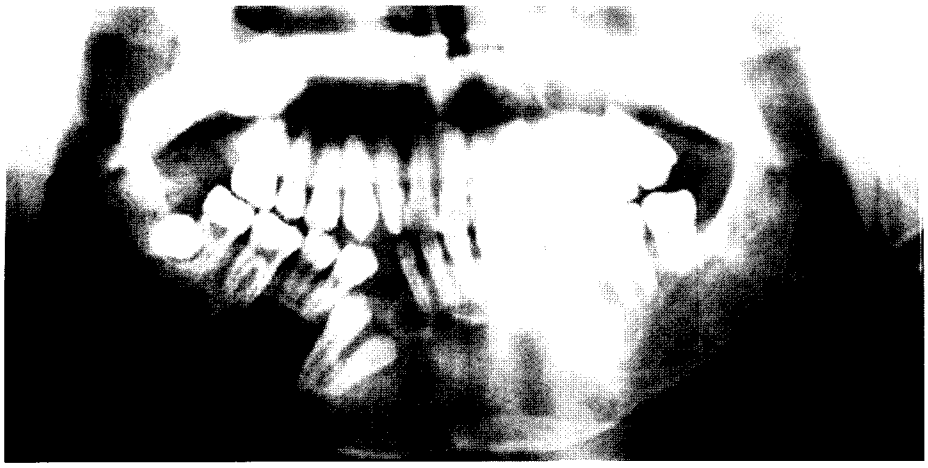

Fig. 1. Well defined radiolucent leions on mandibular anterior region, right and left mandibular posterior area, and left maxillary posterior area.

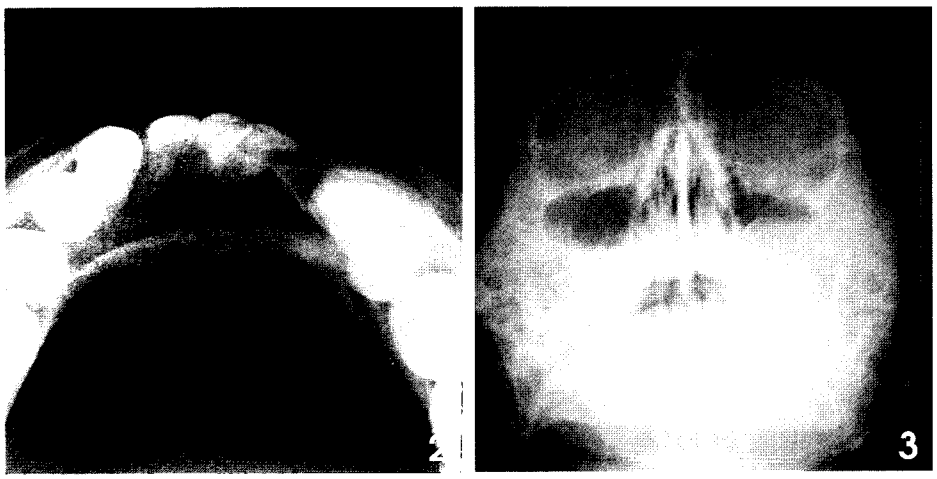

Fig. 2. Cortical expansion and thinning on lower anterior lesion.
Fig. 3. Dome shaped radiopacity with well defined border of left maxillary sinus.

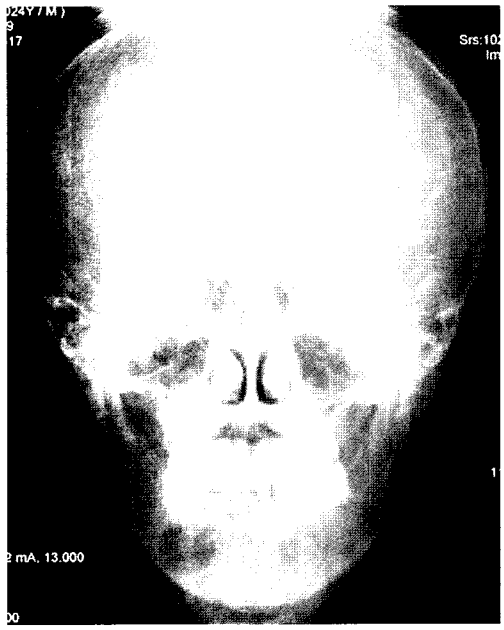

Fig. 4. Calcifications of the falx cerebri and parietal bossing were evident on skull PA view.

q31) was reported as causative gene of this disorder and this gene was believed to act as tumor suppressor gene.[2]

Classic BCNS consists of multiple basal cell carcinoma, multiple odontogenic keratocyst, and skeletal abnormalities and lamellar falx calcification.[14] Recent studies have greatly expanded the syndrome complex to include a variety of other abnormalities.[15-18] Less frequent abnormalities were frontal and parietal bossing, high-arched palate, cleft palate and lip, hypertelorism, congenital blindness, mental retardation, medulloblastoma, ovarian cysts, hypogonadism etc. The basal cell carcinomas mostly affect thoracic and cervico-facial skin surfaces,[13] varying from light to brown dark papules.[19] Palmar and plantar pits are present in 50% to 65% of BCNS patients[20] and are strong indicators of the syndrome.[14,21] The pits are usually 1 to 3 mm in depth and 2 to 3 mm in diameter.[22] In our report, facial nevus was manifested as dermatological abnormalities. Falx cerebri and mental retardation was also observed, but we cannot confirm any familial history. Chenevix-Trench et al.[23] stated that calcification of the falx cerebri was found in

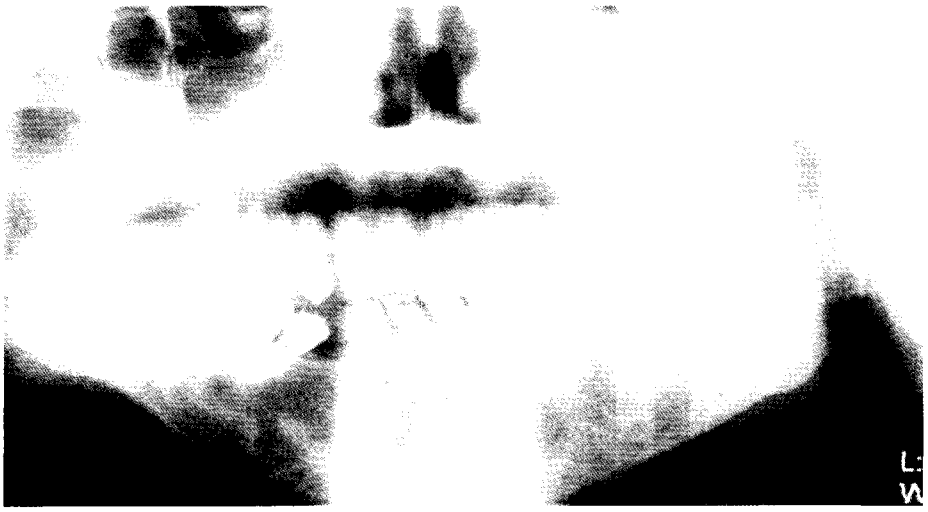

Fig. 5. Reduced radiolucency of pre -cystic lesions after operation.

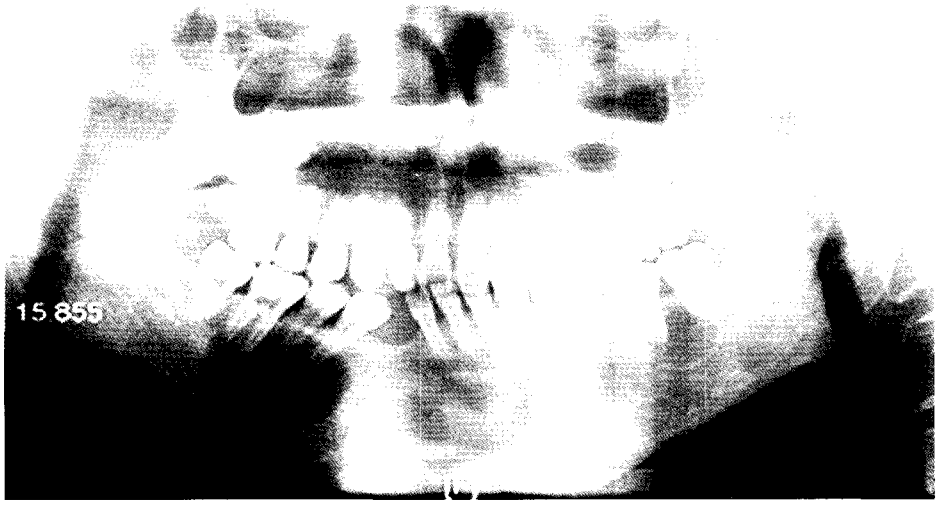

Fig. 6. Newly developed and recurrent cystic lesions on right and left lower posterior area.

Fig. 7. Well defined radiolucent cystic lesion on left mandibular posterior area.

85% to 90% of BCNS. Lovin et al. reported that up to one-third of cases do not have a positive family history.[24]

The OKC has been reported as most frequent characteristics of basal cell nevus syndrome.[9,25] Several workers suggested that half of multiple OKCs involve BCNS.[26] In patients with BCNS, 65%[27] to 100%[28] have OKC. The high recurrence rate of OKC associated with basal cell nevus syndrome. This recurrence rate 6% to 60%.[25,29] It is most likely to occur in the first

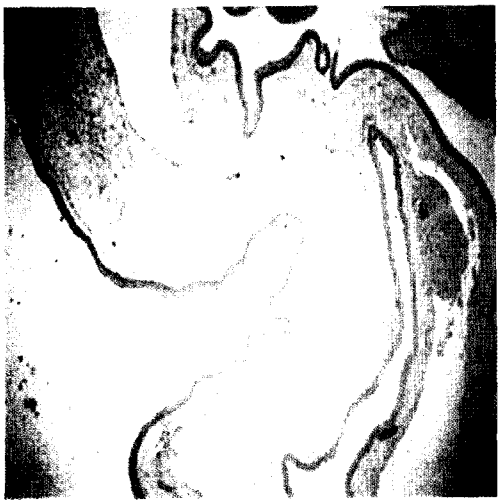

Fig. 8. Typical appearance of odontogenic keratocyst, such as thin epithelium, a well defined basal cell layer and a lumen containing desquamated keratin (H & E stain, × 100).

5 postopertive years.[25]

It has been suggested that recurrence rate of OKC in patients with BCNS may be higher than of OKC in healthy subjects, probably as these former patients have an inherited tendency within the basal layer of their epithelium to develop new cysts.[25,30]

Payne[31] and Stoelinga et al.[32] reported that the recurring tendency is thought to be correlated to the occurrence of microcysts and epithelial remnants in the cyst wall. Browne[33] found that there were no significant difference in the recurrence rate following treatment by three basic methods: marsupialization, enucleation and primary closure. He concluded that high rate of recurrence was related to the nature of the lesion and not to the method of treatment. Brannon[34] and Wright[35] suggested the interesting report that cysts with parakeratinization have a greater tendency for recurrence and another report[36] that OKC may be unilocular in smaller lesions but tends towards multilocularity in larger lesions, a single keratocyst is the lower end of the spectrum of BCNS.[37]

The cysts vary in size and number, and may develop in the maxilla or mandible. The majority of cysts occur in the mandibular molar and ramus regions.[36] Kimonis et al.[38] reported that males and females are equally affected, the clinical features of BCNS arising in the first, second or third decade. Lo Muzio et al.[19] suggested that OKCs arise earlier in patients who have BCNS than in patients who do not have BCNS.

Our patient represented multiple OKC, facial nevus, calcification of falx cerebri, parietal bossing and mental retardation. The cyst on posterior mandibular jaw showed recurrent and newly developing tendency. We diagnosed this case as basal cell nevus syndrome with recurrent OKC. We have made treatment and periodic follow-up check of jaw cystic lesions.

References

1. Bialer MG, Gailani MR, McLaughlin JA, Petrikovsky B, Bale AE. Prenatal diagnosis of Gorlin syndrome. Lancet 1994; 344 : 477.
2. Bonifas JM, Bare JW, Kerschmann RL, Master SP, Epstein EH, Jr. Parental origin of chromosome 9q22.3-q31 lost in basal cell carcinomas from basal cell nevus syndrome patients. Hum Mol Genet 1994; 3 : 447-8.
3. Jarish W. Zur lehre von den autgeschwulsten. Archiv Jur Dermatologic und Syphilogic 1894; 28 : 163-222.
4. Howell JB, Caro MR. The basal-cell nevus: its relationship to multiple cutaneous cancers and associated anomalies of development. AMA Arch Derm 1959; 79 : 67-77; discussion 77-80.
5. Gorlin RJ, Goltz RW. Multiple nevoid basal-cell epithelioma, jaw cysts and bifid rib. A syndrome. N Engl J Med 1960; 262 : 908-12.
6. Gorlin RJ. Nevoid basal cell carcinoma syndrome. Dermatol Clin 1995; 13 : 113-25.
7. Woolgar JA, Rippin JW, Browne RM. The odontogenic keratocyst and its occurrence in the nevoid basal cell carcinoma syndrome. Oral Surg Oral Med Oral Pathol 1987; 64 : 727-30.
8. Gorlin RJ, Pindgorg JJ, Cohen MM. Multiple nevoid basal carcinoma syndrome. In: Gorlin RJ, ed. Syndromes of head and neck. 2nd ed. New York: McGraw-Hill, 1976; 520-6.
9. Mustaciuolo VW, Brahney CP, Aria AA. Recurrent keratocysts in basal cell nevus syndrome: review of the literature and report of a case. J Oral Maxillofac Surg 1989; 47 : 870-3.
10. Howell JB, Byrd L, McClendon JL, Anderson DE. Identification and treatment of jaw cysts in the nevoid basal cell carcinoma syndrome. J Oral Surg 1967; 25 : 129-38.
11. Anderson DE, Cook WA. Jaw cysts and the basal cell nevus syndrome. J Oral Surg 1966; 24 : 15-26.
12. Gorlin RJ. Nevoid basal cell carcinoma (Gorlin) syndrome: unanswered issues. J Lab Clin Med 1999; 134 : 551-2.
13. Gorlin RJ. Nevoid basal-cell carcinoma syndrome. Medicine (Baltimore) 1987; 66 : 98-113.
14. Gorlin RJ, Sedano HO. The multiple nevoid basal cell carcinoma syndrome revisited. Birth Defects Orig Artic Ser 1971; 7 : 140-8.
15. Kopp WK, Klatell J, Blake M. Basal-cell nevus syndrome with other abnormalities. Report of a case. Oral Surg Oral Med Oral Pathol 1969; 27 : 9-14.
16. Ellis DJ, Akin RK, Bernhard R. Nevoid basal cell carcinoma syndrome: report of case. J Oral Surg 1972; 30 : 851-6.
17. van Dijk E, Neering H. The association of cleft lip and palate with basal cell nevus syndrome. Oral Surg Oral Med Oral Pathol 1980; 50 : 214-6.
18. Ruprecht A, Austermann KH, Umstadt H. Cleft lip and palate, seldom seen features of the Gorlin-Goltz syndrome. Dentomaxillofac Radiol 1987; 16 : 99-103.
19. Lo Muzio L, Nocini P, Bucci P, Pannone G, Consolo U, Procaccini M. Early diagnosis of nevoid basal cell carcinoma syndrome. J Am Dent Assoc 1999; 130 : 669-74.
20. Esposito SJ, Kast G, Bradrick JP. Basal cell nevus syndrome: a clinical report. J Prosthet Dent 1995; 73 : 405-10.

21. Potaznik D, Steinherz P. Multiple nevoid basal cell carcinoma syndrome and Hodgkin's disease. Cancer 1984; 53 : 2713-5.
22. Friedlander AH, Herbosa EG, Peoples JR, 3rd. Ocular hypertelorism, facial basal cell carcinomas, and multiple odontogenic keratocysts of the jaws. J Am Dent Assoc 1988; 116 : 887-9.
23. Chenevix-Trench G, Wicking C, Berkman J, Sharpe H, Hockey A, Haan E, et al. Further localization of the gene for nevoid basal cell carcinoma syndrome (NBCCS) in 15 Australasian families: linkage and loss of heterozygosity. Am J Hum Genet 1993; 53 : 760-7.
24. Lovin JD, Talarico CL, Wegert SL, Gaynor LF, Sutley SS. Gorlin's syndrome with associated odontogenic cysts. Pediatr Radiol 1991; 21 : 584-7.
25. Donatsky O, Hjorting-Hansen E. Recurrence of the odontogenic keratocyst in 13 patients with the nevoid basal cell carcinoma syndrome. A 6-year follow-up. Int J Oral Surg 1980; 9 : 173-9.
26. Brannon RB. The odontogenic keratocyst. A clinicopathologic study of 312 cases. Part I. Clinical features. Oral Surg Oral Med Oral Pathol 1976; 42 : 54-72.
27. Southwick GJ, Schwartz RA. The basal cell nevus syndrome: disasters occurring among a series of 36 patients. Cancer 1979; 44 : 22 94-305.
28. Littler BO. Gorlin's syndrome and the heart. Br J Oral Surg 1979; 17 : 135-46.
29. Vedtofte P, Praetorius F. Recurrence of the odontogenic keratocyst in relation to clinical and histological features. A 20-year follow-up study of 72 patients. Int J Oral Surg 1979; 8 : 412-20.
30. Woolgar JA, Rippin JW, Browne RM. A comparative histological study of odontogenic keratocysts in basal cell nevus syndrome and control patients. J Oral Pathol 1987; 16 : 75-80.
31. Payne TF. An analysis of the clinical and histopathologic parameters of the odontogenic keratocyst. Oral Surg Oral Med Oral Pathol 1972; 33 : 538-46.
32. Stoelinga PJ, Cohen MM Jr, Morgan AF. The origin of keratocysts in the basal cell nevus syndrome. J Oral Surg 1975; 33 : 659-63.
33. Browne RM. The odontogenic keratocyst. Clinical aspects. Br Dent J 1970; 128 : 225-31.
34. Brannon RB. The odontogenic keratocyst. A clinicopathologic study of 312 cases. Part II. Histologic features. Oral Surg Oral Med Oral Pathol 1977; 43 : 233-55.
35. Wright JM. The odontogenic keratocyst: orthokeratinized variant. Oral Surg Oral Med Oral Pathol 1981; 51 : 609-18.
36. Kakarantza-Angelopoulou E, Nicolatou O. Odontogenic keratocysts: clinicopathologic study of 87 cases. J Oral Maxillofac Surg 1990; 48 : 593-9; discussion 99-600.
37. Browne RM. The odontogenic keratocyst. Histological features and their correlation with clinical behaviour. Br Dent J 1971; 131 : 249-59.
38. Kimonis VE, Goldstein AM, Pastakia B, Yang ML, Kase R, DiGiovanna JJ, et al. Clinical manifestations in 105 persons with nevoid basal cell carcinoma syndrome. Am J Med Genet 1997; 69 : 2 99-308.