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Case report

Familial occurrence of periapical cemental dysplasia

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Abstract. A family with periapical cemental dysplasia is reported. The affected individuals displayed classical features of periapical cemental dysplasia on radiographic examination. The lesions consisted chiefly of radiolucent areas; however, some had central areas of radiodensity. Histopathological examination of one of the lesions revealed fibrous elements containing fused dense sclerotic cemental masses. Familial incidence of florid cemento-osseous dysplasia with an autosomal mode of inheritance has been reported previously. The condition described in this report appears to be different. However, the two conditions may be part of a spectrum occurring in a single genetic entity with the diversity possibly resulting from variable expressivity of a single gene.

Key words: Periapical cemental dysplasia – Familial incidence

Introduction

Cementomas are a heterogeneous group of lesions affecting the tooth-bearing regions of the jaw. They can be classified into four major types according to the WHO Classification (Kramer et al. 1992). These include cemento-ossifying fibroma, the benign cementoblastoma and the two cemento-osseous dysplasias, periapical cemental dysplasia (PCD) and florid cemento-osseous dysplasia (FCOD). This classification is based on the sex and age of the affected individuals together with the site, radiographic and histological features of the lesions. However, lesions are not always typical and then cannot be satisfactorily assigned to a particular category. Of these lesions, FCOD (formerly termed gigantiform cementoma) is thought to have a familial basis, although until recently only a single family had been described with this condition (Agazzi and Belloni 1953). However, a large pedigree with FCOD has recently been reported (Young et al. 1989). FCOD is regarded as a rare benign condition in which the lesions are often multiple and symmetrical in their distribution. It may present as an incidental finding on radiographic examination or as a bony hard swelling of the jaws. FCOD is thought to occur predominantly in middle-aged women of Afro-Caribbean origin. However, it is a relatively rare condition and only a few proven cases have been reported (Agazzi and Belloni 1953; Gorlin et al. 1961; Lyons and Babajews 1986; Punniamoorthy 1980; Van der Waal and Van der Kwast 1974; Winer et al. 1972; Zegarelli et al. 1964). Histologically, FCODs consist of large sheets or fused globules of cemento-osseous tissue and this is reflected in the radiographic findings of radiodense lobulated masses (Kramer et al. 1992). In contrast, PCD is restricted to periapical tissues predominantly affecting post-menopausal females and is commonly localized to the mandibular incisor region (Kramer et al. 1992). The condition is usually symptomless and often discovered as an incidental finding on radiographs. The lesions are associated with vital teeth. Based on radiographic and histological features, three stages are recognized in the development of PCD (Makek 1983); an early osteolytic stage, an intermediate cementoblastic stage and a mature inactive stage. The early stage is characterized by replacement of periapical bony tissue by fibrous tissue and radiographically appears as apical radiolucent areas similar to those observed in periapical inflammatory disease. The cementoblastic stage is characterized by formation of cementicles which fuse to form larger masses of cementum. In the central areas, calcifying woven tissue is often observed. Radiographically, radiodense areas are seen in the centre of the radiolucent areas. In the final inactive stage, the lesion appears as apical radiodense masses with a radiolucent border and histologically consists mainly of acellular cementum. In this paper, we report a family showing classical features of PCD. Familial cases of this condition have not been reported previously.

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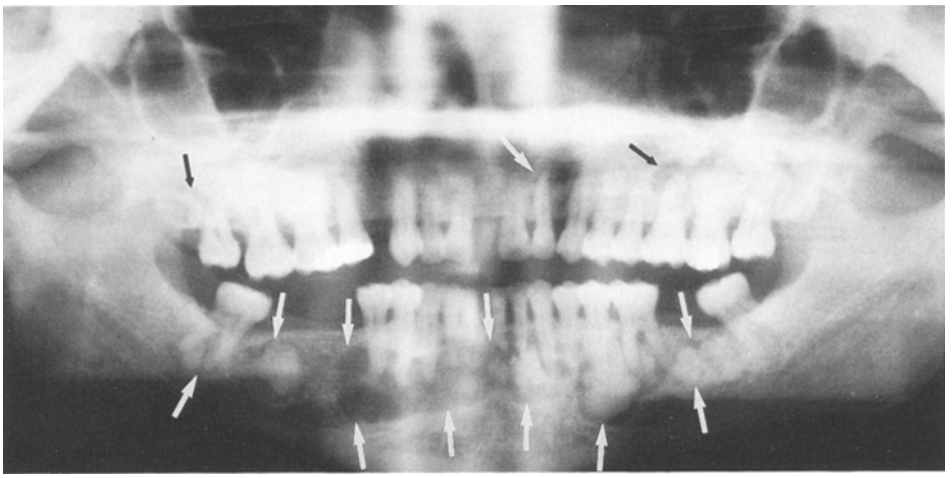

Fig. 1. Orthopantomograph of the proband showing multiple periapical radiolucent areas with central radiodense regions typical of periapical cemental dysplasia (*arrows*)

Case report

A 61-year-old Afro-Caribbean woman was referred to the Department of Oral and Maxillofacial Medicine and Surgery for an opinion by her general dental practitioner regarding discomfort from, and mobility of, mandibular incisors associated with multiple periapical radiolucent areas. The symptoms had been present for a few weeks. Her past medical history was unremarkable with no evidence of any systemic disease. On examination, there was no expansion or palpable abnormality of the jaws; the teeth were not carious and electrical vitality tests were positive. However, there was some gingival recession around the mandibular incisors due to chronic periodontal disease. Periapical radiographs of these teeth showed periapical radiolucent areas with central radiodense regions. Subsequent orthopantomography revealed similar lesions associated with numerous teeth in both the maxilla and the mandible (Fig. 1). The symptoms relating to the mandibular incisors were attributed to chronic periodontal disease and necessitated the extraction of three teeth. Subsequent socket healing was uneventful. The extracted teeth and associated periapical tissues were demineralized and processed for routine histological examination. The teeth showed normal dentine structure with vital pulp. The apical tissue consisted of fibrous elements containing fused dense sclerotic cemento-osseous masses (Fig. 2). With regard to the radiographic anomalies, a diagnosis of PCD was made. In order to elucidate a possible familial basis for this condition and its relationship with FCOD, it was decided to investigate other members of the family. The family pedigree is shown in Fig. 3. Several members of the family (indicated by an asterix) were not available for examination. Four of the seven children of the proband were examined. Three of the children had jaw lesions similar to those observed in the proband. These were a son aged 34 (Fig. 4) and two daughters aged 30 (Fig. 5) and 26 (Fig. 6). All the affected children had lesions predominantly in the mandibular incisor region although early small lesions were seen associated with apices of other teeth in both the jaws. The lesions consisted chiefly of radiolucent areas; however, some had central areas of radiodensity (Fig. 7). All radiographs were independently assessed by a consultant radiologist who reported that the features were typical of PCD.

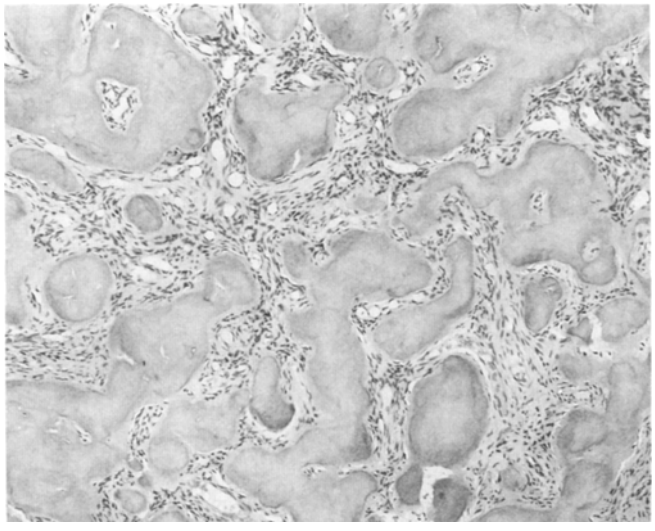

Fig. 2. Photomicrograph showing dense fused globular cemento-osseous masses in a fibrous matrix. Haematoxylin and eosin ×150

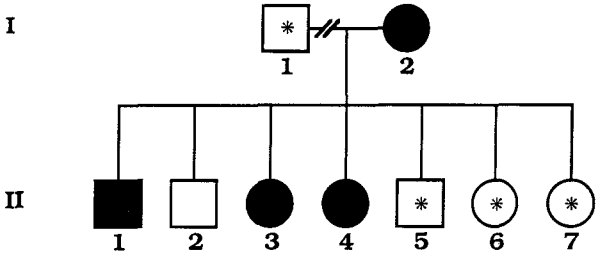

Fig. 3. Pedigree of the PCD family. Individuals indicated by asterix were not available for examination

Discussion

The family presented in this paper shows features typically associated with PCD. This condition is thought to be self-limiting and restricted to the periapical tissues. Familial cases have not been described previously. In contrast, FCOD is thought to have a familial basis (Agazzi and Belloni 1953; Melrose et al. 1976) although only one well-characterized pedigree has been reported so far (Young et al. 1989). The previous lack of evidence for familial incidence of PCD may simply be due to the asymptomatic nature of the condition and failure to examine families of patients with PCD. Individuals with FCOD are more likely to present clinically because

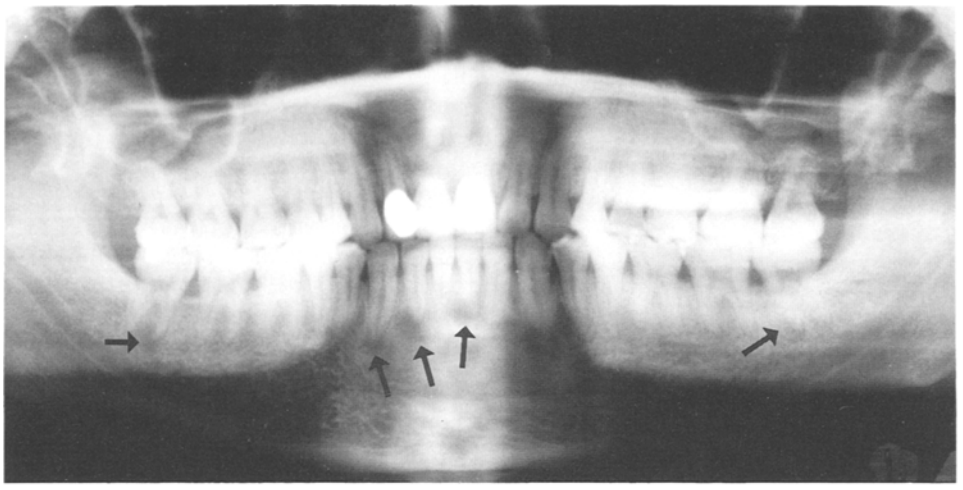

Fig. 4. Orthopantomograph of the probands son (II-2, Fig. 3) showing multiple periapical radiolucent lesions (*arrows*), mostly in the lower incisor region

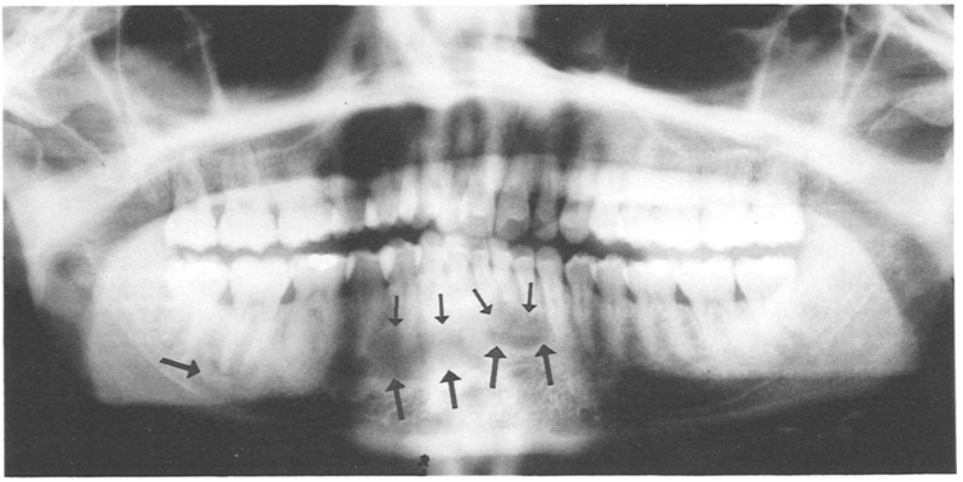

Fig. 5. Orthopantomograph of the probands elder daughter (II-3, Fig. 3) showing periapical radiolucent and radiodense lesions (*arrows*)

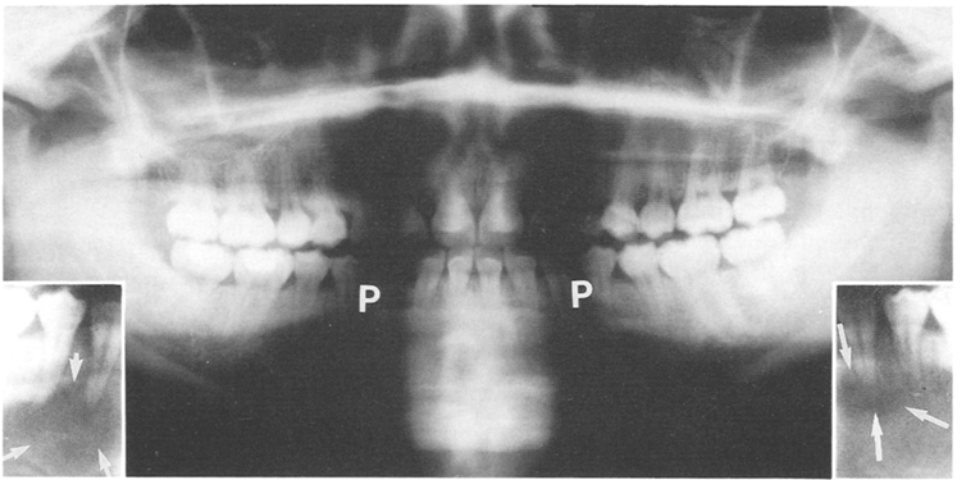

Fig. 6. Orthopantomograph of the probands younger daughter (II-4, Fig. 3). The periapical regions of most teeth were unaffected. Insets show the mandibular premolar regions (*P*), where periapical radiolucencies (*arrows*) were present in relation to the first premolars (original radiograph was high contrast)

cortical expansion often occurs in this condition. Since we were unable to examine the proband's other children, siblings, parents and husband, we are unable to state with confidence the inheritance of PCD in this family. However, since the reported incidence of PCD in the general population is 2–3/1000 (Zegarelli et al. 1964), it is unlikely that four individuals with the disease would be observed in the same family. Furthermore, the disease is more common in females than males and in the family reported here, one male is affected. Accepting a familial basis for PCD in this family, the likely mode of inheritance is autosomal dominant. However, we cannot exclude environmental factors or other modes of inheritance without further data. With regards to the relation-

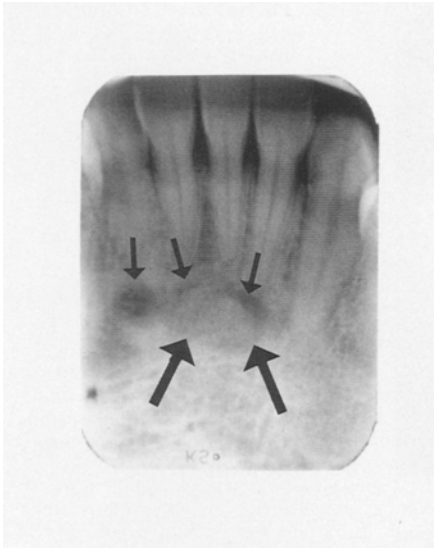

Fig. 7. Periapical radiograph of one of the lesions from individual II-3 (Fig. 3) showing in greater detail the apical radiolucency with central radiodense areas (*arrows*)

ship of PCD to FCOD, several differences do exist between the lesions in the family reported here and those in the only well-characterized FCOD family reported so far (Young et al. 1989). The FCODs often presented early (youngest patient being 6-years-old) with a massive expansion of the jaws that required surgical correction. The age of the proband in the family reported here was 61 years. Furthermore, the lesions were asymptomatic and only discovered on radiographic examination. Thus, it is difficult to estimate the age at which these lesions occurred. The radiographic features of the FCOD lesions were described as "extensive radiopaque areas" and were clearly unlike the periapical radiopacities with areas of radiodensity seen in our patients. Thus it appears that there may be two distinct types of "familial cementomas". However, the classification of cemental lesions (based on the sex and age of affected individuals together with the site, radiographic and histological features of the lesions) though useful, is often difficult and inaccurate. Lesions are often observed that cannot be satisfactorily designated to a particular category. It may be that the lesions observed in the family reported here and in familial FCOD are part of a spectrum occurring in a single genetic entity. The histogenic evolution of all these lesions may follow that described for PCD i.e. an early fibrous stage followed by increasing calcification eventually resulting in an almost solid acellular cemental mass characteristic of FCOD. This is suggested by the histological features described by Young et al. (1989) in their FCOD family. The lesions were characterized by proliferation of fibrous tissue in which cementicles were observed. In a few areas, the calcified masses fused to form more solid areas. Furthermore, clinically, as in the present family, not all affected individuals in the FCOD family had cortical expansion. The differences in extent and severity of the disease may be attributable to variable expressivity of a single gene or due to different rates of evolution of the lesions. This is suggested by the more extensive lesions seen in the proband in the family reported here compared to her affected children. Previous studies of apparently sporadic FCODs also have alluded to the notion that both FCOD and PCD are part of the spectrum of the same benign cemento-osseous condition (Melrose et al. 1976; Waldron et al. 1975). Both conditions have been reported to occur more frequently in Afro-Caribbean females (although none of the previously reported familial cases of FCOD have been in Afro-Caribbeans). The condition reported here is benign and does not warrant treatment. Even in classical FCOD, treatment is only indicated to correct facial deformity. However, both PCD and FCOD may be confused with apical pathosis and may complicate dental extractions. The value of screening in these conditions is to establish correct diagnosis and prevent unnecessary treatment or complications in dental management.

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