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CASE reports

Chondrodysplasia punctata: case report with dental findings

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Chondrodysplasia punctata is a rare growth disorder characterized by ectopic calcification of cartilage. This disease was described first by Conradi in 1914.[1] Since that time this disease has been described under several different names including chondrodystrophia calcificans congenita punctata, chondroangiopathia calcanea seu punctata, chondrodystrophia calcificans congenita, congenital stippled epiphyses, epiphyseal dysplasia, and metaphyseal dyscrasia.[2-7]

This disease is classified into 2 phenotypes and their differentiation is based on the mode of inheritance as well as clinical findings (Table). The first is Conradi-Hunermann type, referred to as Conradi's disease, which is inherited as an autosomal dominant trait.[8] The prognosis with this type is good if the patient survives the first 3 months.[9,10] The second phenotype is rhizomelic variety, inherited as an autosomal recessive trait, in which survival past the first year is poor.

Chondrodysplasia punctata has not been reported previously in the dental literature. The purpose of this report is to present a case of Conradi's disease with interesting dental findings.

Chondrodysplasia Punctata

The diagnosis of this disease is based on the radiographic findings at birth. Affected infants show minute punctate calcifications primarily in the epiphyseal cartilages of long bones. Calcifications also can be observed in other cartilagenous areas of the newborn such as the ribs, sternum, larynx, vertebrae, and ischiopubic bones. Usually these calcifications disappear toward the end of the first year of life, making the definitive diagnosis of this disease difficult after the first year. Also confusing the diagnosis is the fact that punctate calcifications may be present in several other disorders including Down's syndrome, trisomy 18, Zellweger syndrome, hypothyroidism, spondyloepiphyseal dysplasia, anencephaly, and cerebrohepatorenal syndrome.[9,10] Furthermore, punctate calcifications may occur as a harmless isolated anomaly.

Common features of this disease include short stature, short neck, congenital cataracts, flexion joint contractures, saddle nose deformity, and numerous dermatoses. Less frequent manifestations may include cardiac malformations, frontal bossing, malar hypoplasia, high-arched palate, and micrognathia.[1,5,11-13] Affected individuals have no higher prevalence of mental retardation and motor retardation is rare.[14] Mild to moderately affected individuals usually survive to adulthood and retain their reproductive capacity.[1,9] Severely affected individuals usually die early in infancy.[1,5,15]

Case Report

Medical History

The patient was a 5-pound, 13-ounce product of an uncomplicated full-term pregnancy which yielded fraternal twins by caesarean section. Chest radiographs were obtained shortly after delivery because of respiratory distress. These radiographs were found to reveal stippled epiphyses and iliac wings. The radiographic features at birth suggested chondrodysplasia punctata; therefore, the infant was referred for examination to the Division of Genetics in the Department of Pediatrics at the North Carolina Memorial Hospital. At 4 months of age she was diag-

TABLE. Differentiation of Phenotypes in Chondrodysplasia Punctata

Physical Features	*Conradi-Hunermann*	*Rhizomelic*
Head	Usually normal but with prominent forehead, flat nose, hypertelorism, and upward palpebral slant	Microcephaly usually present
Eyes	Cataracts ~ 20%	Cataracts ~ 75%
Limbs	Large joint contractures ~ 25%	Severe foreshortening of humeri and femora
Skin	Atrophoderma follicularis is frequent, cicatricial alopecia occurs often	No changes reported
Height	Newborn is short, adult height from 130 cm to normal	Mean birth length: 48.2 cm Linear growth is usually below normal
Mental retardation	Rarely present	Usually present
Genetics	Dominant trait	Recessive trait
Radiologic findings	Absent	Present
Severe bilateral shortening of femora and humeri		
Severe metaphyseal changes	Absent	Present
Distribution of lesions	Frequently asymmetric	Usually symmetric
Vertebral column	Scoliosis often present	Coronal cleft in lateral aspects of vertebral bodies
Prognosis	If infant survives first 3 months, survival to adulthood is likely	No patient has survived to adolescence

Modified from Louvar RD, Block WM, Martin LR[15]

nosed as having chondrodysplasia punctata, Conradi-Hunermann type. The twin brother was examined and exhibited none of the clinical or radiographic findings consistent with the disease.

The clinical and radiographic findings of this case followed a course consistent with chondrodysplasia punctata. Stippling of vertebral bodies T-7 to T-11 was noted at birth. There was bilateral ptosis and depression of the nasal bridge. Stippling of the vertebral bodies disappeared by age 1, but there was dorsal kyphosis in the area of the previous stippling. Additionally, stippling was noted on the left iliac crest. The kyphoscoliosis was stable until approximately age 6. At that point the scoliosis was found to have increased over the previous 2 years.

Ear infections during the first year led to myringotomy and tube placement at age 2. The child's surgical history included tonsillectomy and adenoidectomy operations at age 4, a Fasanella-Servat operation on her right eye at age 4, and surgical repair of bilateral congenital ptosis at age 7. The patient was being followed for possible orthopedic correction of her kyphoscoliosis. Her height was at the 90th percentile and weight was at the 80th percentile for her age. She was of normal intelligence and was progressing well in the appropriate school grade.

Dental Findings

The patient was a 7-year-old black female referred to the Department of Pediatric Dentistry with a chief complaint of "enamel missing on front teeth." There had been no previous dental treatment.

The extraoral examination revealed a symmetrical face, but there was bilateral ptosis and a depressed nasal bridge. In profile, bimaxillary protrusion and an apparent mandibular prognathism were noted. No oral habits were reported. Two round nodules were present on the lower lip; each was ~ 4 mm in diameter and slightly elevated. The parents reported that since their first appearance at age 1, there had been no change in size, color, or texture in these nodules. A tentative diagnosis of benign nevi was made.

Intraoral findings revealed a mixed dentition with a Class I occlusal relationship for both permanent molars and primary canines. There was a dentoalveolar open bite of 7 mm. The lower primary first molars appeared to be ankylosed. Dental caries was limited to pits and fissures of the posterior teeth. Radiographic analysis revealed the presence of all developing succedaneous teeth as well as developing second and third permanent molars. Radiographically, generalized enamel hypoplasia of the erupted dentition could be seen and there was apparent hypoplasia of the crowns of the unerupted premolars and molars. (Figs 1, 2)

Treatment included instruction in proper oral self-care, alloy restorations for the decayed pits and fissures of the posterior teeth, sealants for the caries-

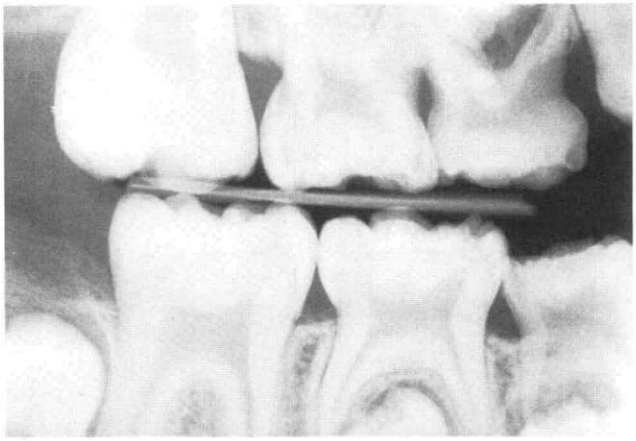

FIG 1. Bite-wing radiograph illustrating enamel hypoplasia affecting the crowns of the primary molars and 6-year molars.

free pits and fissures of the posterior teeth, sealants for the hypoplastic areas on the facial surfaces of the mandibular incisors, interim resin restorations for the maxillary incisors, and consultation with the Department of Oral Pathology regarding the lower lip lesions.

The oral pathologist diagnosed the lip lesions to be benign nevi and felt that their removal was elective. The patient and her parents decided against removal because the lesions were not esthetically displeasing to them.

At the time of the diagnosis of chondrodysplasia punctata, the parents received extensive genetic counseling regarding the transmission of the disease. Furthermore, they were aware that their child was a carrier of this autosomal dominant disease trait and the parents planned to have genetic counseling for her at adolescence.

Discussion

Chondrodysplasia punctata is a rare, congenital developmental disorder and patients affected with the less severe form may be expected to live normal, reproductive lives. Because the physical features of these patients are unusual and may appear similar to other craniofacial anomalies or syndromes such as Down's, the patients are likely to present to the pediatric dentist by referral. The pediatric dentist should be aware of several factors in dealing with such patients and families.

1. A genetic consultation is advisable for parents of a child who presents with multiple dysmorphic features with no definitive diagnosis.
2. With early intervention and proper medical management, the physical features of the patient with chondrodysplasia punctata can be addressed.
3. Generalized enamel hypoplasia in both dentitions may be a feature of this disease.

The patient presented with clinical and radiographic findings consistent with Conradi-Hunermann variety of chondrodysplasia punctata. Her developmental course has followed that expected with this disease including the disappearance of vertebral stippling in the first year of life and the appearance of scoliosis in early childhood. Her height, weight, and cognitive development were age appropriate.

The pattern of enamel hypoplasia in this patient confirms that the etiology was unrelated to a single isolated event; rather, the causative factor extended over a period of years or resulted from a systemic problem. Although chronic ear infections and their concomitant fevers and medications may be correlated with enamel hypoplasia, the ear infections between age 1 and 2 were not sufficient to explain hypoplasia in this case. In the absence of a definitive etiology for the enamel hypoplasia, it could be speculated that it was present in this patient in association with the chondrodysplasia punctata.

The defects seen in chondrodysplasia punctata are due to variations in bone formation during endochondral ossification. Among the tissues involved in

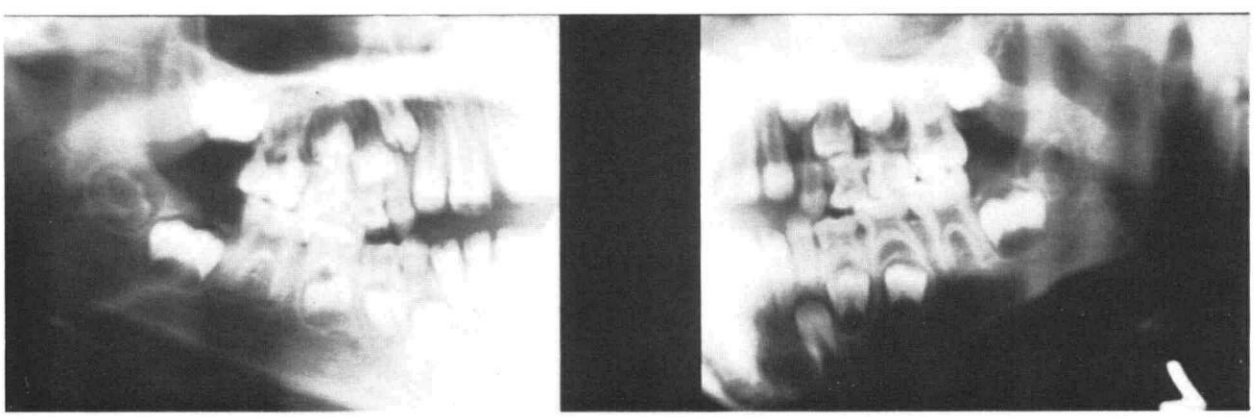

FIG 2. Panorex radiograph illustrating enamel hypoplasia on erupted and unerupted teeth.

this process is cartilage, of which collagen fibers serve as the basic matrix origin.[16] Also, collagen fibers are involved with dentin formation and the presence of dentin is necessary for enamel matrix formation to begin.[17] Therefore, it is possible that if there were a flaw in collagen genesis, a portion of the dentin formed might be defective, and consequently so could its corresponding enamel. Lending credence to this theory (in this patient) is the fact that the enamel defects are dispersed throughout the primary and permanent dentitions without any other identifiable etiology.

The authors recognize that there is a lack of basic biological information tying chondrodysplasia punctata and enamel hypoplasia together. Because the dental findings of chondrodysplasia punctata have not been reported in the medical literature and there are no reports in the dental literature, their association in this case could be coincidental, or they may be related by the mechanism noted here. This report should encourage closer attention to the dental findings in patients with chondrodysplasia punctata.

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National Children's Dental Health Month Art Contest Winners

Entries were received from all parts of the United States and from South America in the cover art contest sponsored by the Academy. The contest, held during February (National Children's Dental Health Month), will be held again in February, 1987.

The winners in this year's contest (followed by the name of their sponsoring dentist) were:

Grand Winner — Diamond Fernando, Sao Paulo, Brazil: Daniel Korytnicki
Runners-Up — Carlos Braun Sampio, Sao Paulo: Daniel Korytnicki; Brian Hammond, Las Cruces, New Mexico: John N. Conniff; Drew Heinerichs, Broomall, Pennsylvania: Sheldon M. Bernick; and Herman Paul Shinkovich, III, Granville, West Virginia: R.E. Day

Many fine entries were received, and each of them was sent a certificate denoting participation.