Trichodental Dysplasia: A Rare Syndrome With Distinct Dental Findings

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Abstract

The association of fine, dry, short hair and the developmental absence of several teeth has been associated with a rare autosomal dominant variant of ectodermal dysplasia known as trichodental syndrome or trichodental dysplasia. The purpose of this study was to present the case of a 4-year-old boy with trichodental syndrome. Clinical, radiographic, and genetic manifestations are described, along with a pertinent review of the literature. (Pediatr Dent 2006;28:345-349)

KEYWORDS: ECTODERMAL DYSPLASIA, TRICHODENTAL, TEETH, GENETIC TESTING, SHELL TEETH

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Congenital absence of 1 or more teeth without any associated anomalies is a rather common condition observed in 4% to 8% of the human population.[1] When other ectodermal derivatives, such as hair, nails, or sweat glands, are simultaneously affected, ectodermal dysplasia emerges as a diagnostic possibility. An extensive review of the congenital disorders—where the disorder affected 2 or more ectodermal derivatives and resulted in ectodermal dysplasia—was conducted by Witkop et al[2] almost 3 decades ago. A variant of ectodermal dysplasia, trichodental syndrome was first described by Salinas and Spector[3] in 1980 and later by Kersey[4] in 1987.

The purpose of this case study was to present the clinical, radiographic, and genetic features of a 4-year-old child with trichodental dysplasia that adds to the existing knowledge of this rare syndrome.[5]

Trichodental syndrome has been described as a rare autosomal dominant condition affecting the hair and teeth. Only a handful of cases have been reported so far. In 1954, Rushton[6] described this form of dentinal dysplasia as teeth with normal enamel and extremely thin dentin with large pulps and short roots. He used the term "shell teeth" to describe the condition. Kersey[4] described a family pedigree having trichodental syndrome with abnormal teeth and fine hair and suggested inheritance may be dominant. Salinas and Spector[3] described the syndrome in 13 patients and determined an association with abnormal hair and hypodontia. Common findings have been fine, lusterless, sparse and slow-growing hair, hypodontia, and/or abnormally developed teeth. Microcephaly and mild mental retardation as symptoms of this rare condition have been added by Giannotti[7] and Kinirons.[8]

Case Report

A 4-year-old Hispanic male presented for a comprehensive examination at the Department of Pediatric Dentistry at the New Jersey Dental School, University of Medicine and Dentistry New Jersey, Newark, NJ. The patient's mother reported him to be in good general health. On examination,

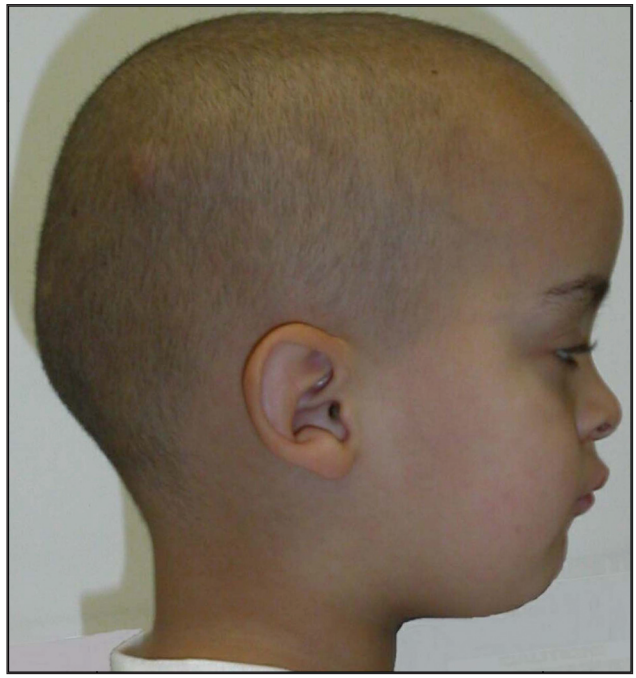

Figure 1. Patient's facial profile. Note the frontal bossing and sparse hair.

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his head circumference was 52 cm (75th percentile for his age group), his height was 105 cm, and he had thin, sparse hair and frontal bossing (Figure 1). His mother reported that a computed tomography (CT) scan was done earlier on the advice of his physician due to concerns about the size and shape of his head and to rule out any deformational plagiocephaly or craniosynostosis. CT scan findings were reported to be within normal limits. Extraoral examination of the patient revealed normal facial features—with somewhat thinner eyebrows, but normal-appearing eyelashes. Facial skin and lips appeared normal, and his mother reported that the child did not have any problems related to perspiration.

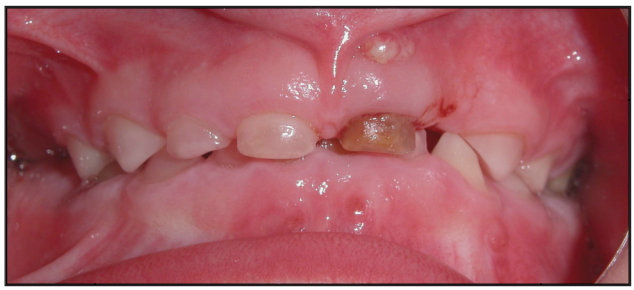

Figure 2. Teeth in occlusion. Note the translucent nature of the teeth and the discolored left maxillary primary central incisor and the associated parulis at the mucogingival junction.

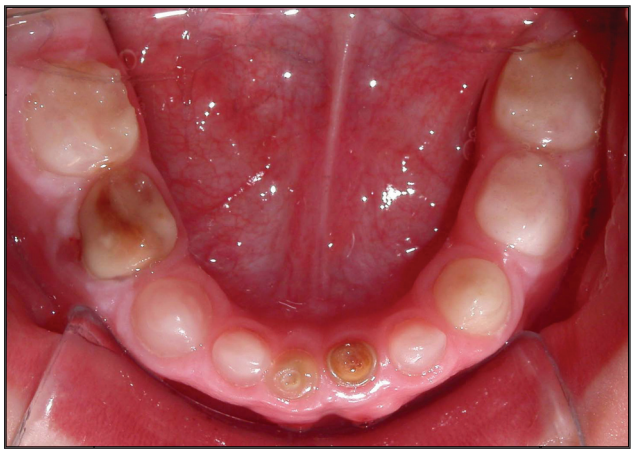

Figure 3. Occlusal view of the mandibular arch.

The intraoral examination revealed a primary dentition with a translucency of all primary teeth. Pulp chambers were visible through the lingual surfaces of the maxillary incisors and occlusal surfaces of the primary molars, representing the thickness of the enamel over these teeth (Figures 2 and 3). The patient had significant attrition on most of the teeth. Dentoalveolar abscesses were noted in 6 of his primary teeth. Intraoral periapical radiographs were taken followed by a panoramic radiograph. It was noticed that all the primary teeth had thin enamel, no visible dentin, and large pulp chambers— giving a "shell-like" form and also exhibiting the typical features of taurodontism (Figures 4 and 5). Furthermore, the developing permanent dentition had similar radiographic morphology, giving the appearance of a generalized variety of odontodysplasia. The patient was referred to a geneticist at the university hospital for a consult and genetic testing to rule out a possible syndrome related to ectodermal dysplasia or a generalized variety of dentinal dysplasia. The geneticist agreed with the provisional diagnosis that the patient's dental anomaly fell into the range of ectodermal dysplasia-related syndromes or generalized odontodysplasia pending further evaluation. After reviewing the patient's history and the reports from the dental examination, the geneticist recommended a cytogenetic analysis of the chromosomes as well as a chromosomal fluorescence in situ hybridization (FISH) test.

There was a history of cleft palate that affected one of the patient's cousins. The geneticist had a concern that the patient may have been somewhat behind developmentally for his age. The fluorescence in situ hybridization (FISH) procedure was performed using a probe for the long arm of chromosome 22 to rule out any additional genetic syndromes. The long arm of chromosome 22 was intact.

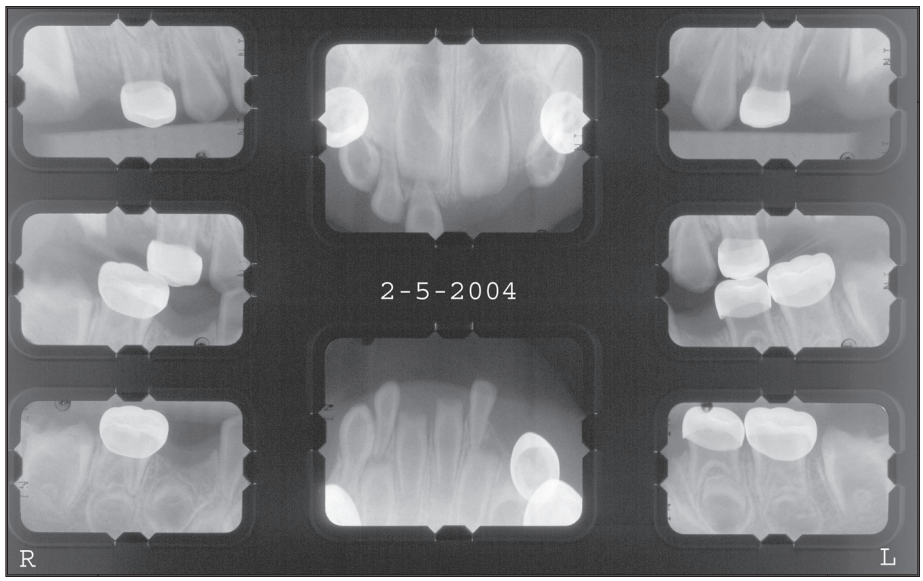

Figure 4. Intraoral periapical and bitewing radiographs of the patient after the second visit. Notice the placement of crowns on all erupted and clinically present primary molars.

The patient had no history of fractures and exhibited normal sweating, but had extremely slow-growing hair, with only 2 prior haircuts since birth. His nails had very fine linear creases with mild fingernail clubbing. The patient's mother was in excellent health, exhibited none of the symptoms that the child demonstrated, and had no history from her side of the family. The patient's father, however, had slow-growing, sparse hair. In addition, the father's 2 sisters expressed thin hair, and one also had missing

teeth. Furthermore, their children had some of these similar findings (Figure 6).

Chromosome (cytogenetic) analysis

Approximately 20 cells were examined, 5 cells were analyzed, and 2 cells were karyotyped with a band resolution of 525. GPW banded metaphases revealed a normal appearing 46,XY male chromosome complement in all cells examined. No numerical or structural chromosomal abnormalities were observed. Considering the paternal history, abnormal hair, missing and/or misshapen teeth, and genetic testing, a provisional diagnosis of trichodental dysplasia was given.

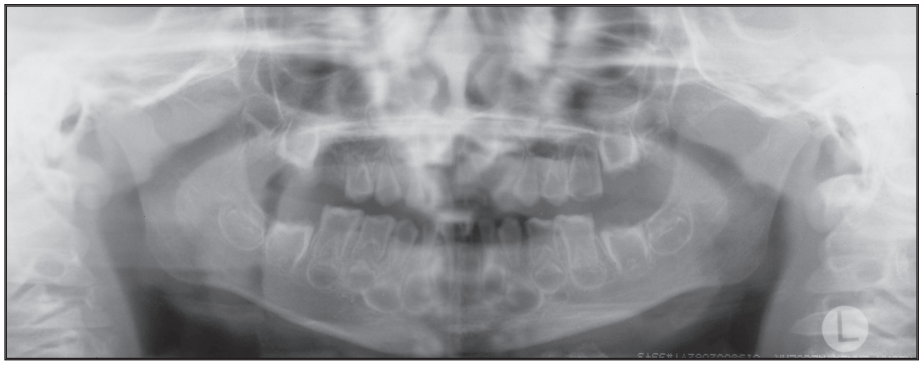

Figure 5. Panoramic radiograph of the patient when presented initially. Both primary and permanent have the appearance of shell teeth and exhibit taurodontism in multirooted teeth.

Treatment plan and follow-up

A treatment plan was completed for the child that included treating the patient in the operating room under general anesthesia. The treatment included extraction of 6 primary teeth due to nonvitality, abscess formation, and nonrestorability. A residual root was removed from the upper right first molar area. Stainless steel crowns were preventively placed on the remaining 5 primary molars without preparing the teeth. The patient's parents were told that crowning the teeth may not be a permanent solution for these teeth and that they may need to be extracted in the future. The remaining anterior teeth were left untreated, as pulp exposure was a concern with acid-etch treatment. Since further carious lesions may result in pulp exposures for this patient, he was placed on frequent recall visits with fluoride treatments as well as daily fluoride regimens.

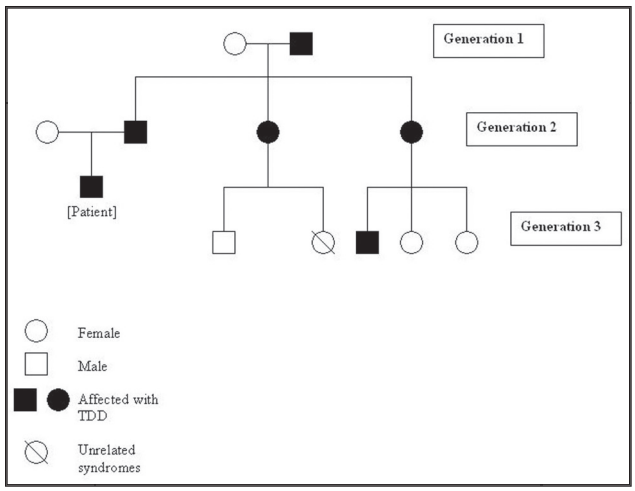

Figure 6. A 3-generation pedigree of the patient generated from the history.

Discussion

Trichodental dysplasia is an autosomal dominant type of ectodermal dysplasia. Ectodermal dysplasias (ED) are a heterogenous group of disorders characterized by developmental dystrophy of ectodermal structures, such as hypohidrosis, hypotrichosis, onychodysplasia, and hypodontia or anodontia. Approximately 160 clinically and genetically distinct hereditary ectodermal dysplasias have been cataloged by Freire-Maia and Pinheiro[9] and were later classified[10] based on their clinical symptoms. Trichodental dysplasia is categorized as a rare disorder, and the clinical manifestations primarily include hair and dental anomalies.

The Christ-Siemens-Touraine (CST) syndrome—often called anhidrotic or hypohidrotic ectodermal dysplasia (AED or HED) because of the inability of affected individuals to sweat normally—is a common suspect in cases where the child presents with congenital absence of teeth, sparse hair, and reduced ability to sweat.[11] Heat intolerance is a frequent complaint. More often, the diagnosis is not made until the teeth do not erupt at the expected age or the teeth appear to be pointed when they do erupt. Eruption may be delayed, or only a few teeth may erupt.[11] Since the aforementioned features were not part of the clinical picture in this case, HED was excluded from the diagnostic possibilities. Moreover, shell teeth were not part of the HED syndrome.

Other variants of ectodermal dysplasia, such as trichodento-osseous (TDO) syndrome and trichorhinophalangeal (TRP) syndrome,[10] closely resemble trichodental dysplasia and should be differentiated based on clinical and radiographic features. Teeth affected by TDO syndrome have thin, pitted, yellow-brown enamel. Teeth may become abscessed during the first few years of life. On intraoral radiographs, large pulp chambers can be found exhibiting taurodontism. In addition, teeth may remain unerupted long past the date when they should be present in the mouth. TDO syndrome patients are found radiographically to have increased density of bone. In some cases, the skull bones are excessively thick. These abnormalities are of no clinical significance and should cause individuals with this syndrome no problems. They are, however, helpful in making the diagnosis. The patient presented did not have abnormal bone density on his radiographs.

Intelligence is normal for TDO syndrome patients, but there may be mild to moderate mental retardation in most affected TRP syndrome patients. Moreover, with TRP syndrome (types 1, 2, or 3), there will be involvement of fingers or toes apart from the changes seen in the hair, teeth, and nails.[11] Patients with TRP syndrome might exhibit multiple bony exostoses as well as chromosomal abnormalities—specifically chromosome 8.[10] The patient had no chromosomal abnormalities or evidence of bony exostosis in the jaws and had mild clubbing of the fingernails. Clubbing is normally seen in disorders of the heart or lungs that limit blood-oxygen levels. Other disorders that could lead to clubbing, such as infective endocarditis, bronchiectasis, cystic fibrosis, lung abscess, chronic liver disease, and celiac disease, were ruled out. TDO and TRP syndromes were also ruled out.

Trichodental dysplasia has been described in a number of families,[3] and the affected family members can have shell teeth, misshaped teeth, hypodontia, or even normal teeth with only typical hair findings. Abnormal hair and missing teeth are the most common manifestations of this autosomal dominant condition, as was discussed by Kersey.[4] The hair in these individuals is described to be fine, sparse, dull, and slow growing. The measured hair growth rate, trichogram, and lack of signs of tip-weathering and trauma suggested that short hair resulted from a short anagen phase of the hair cycle, rather than slow growth.[6] Developmental delays are seen in children diagnosed with this condition.

In the report presented by Giannotti and colleagues,[7] their patient had mild mental retardation with borderline microcephaly. Teeth in general resemble those seen in regional odontodysplasia (ghost teeth). Hence, some of the cases have been reported primarily as "generalized shell teeth" or "generalized odontodysplasia." The teeth in these reports had extremely thin enamel and dentin and enormous pulp chambers.[8,12] Typically, the condition affects a focal area of dentition,[1] although there have been reports of generalized occurrences that are nonsyndromic in occurrence.[13,14]

The cases resemble a condition, now known as dentinogenesis imperfecta type 3 (Shields type III). The condition was also previously referred to in the literature as focal odontoblastic dysplasia by Eastman, et al.[15]

Shell teeth was a common finding in separate cases reported both by Rushton[6] (1954) and Kinirons[8] (1984). Kinirons reported that the patient presented had blue sclera and a history of fractured long bones. He concluded that findings were consistent with an extreme variant of dentinogenesis imperfecta. Rushton[6] described the clinical findings in an adult, whereas Kinirons'[8] subject was an 8-year-old boy.

Intrafamilial and interfamilial variation is expected in many disorders due to autosomal dominant mutations. This seems to be the case in trichodental syndrome, where the pattern of hair and dental abnormalities are rather wide.[7] A tendency towards premature balding has been reported in some patients, although the most significant anomalies of hair were fine, straight, dull, and lusterless appearance. According to Giannotti and associaties,[7] gross examination of the hair in adult patients revealed a large proportion of white hair and a slight beading effect, resulting from variation in reflectance along individual hair shafts. Microscopic examination in their report revealed thin shafts of hair, beading effect related to variations in shaft contour, and alterations in the cuticular pattern including lack of cuticle or reduced scaling. Intraoral examination among these patients revealed a wide range of tooth abnormalities as well. Thinning of the lateral ends of the eyebrows was another interesting feature described in the report by Salinas and Spector.[3]

Kamen and associates[16] have described a condition that they referred to as shell teeth (Brandywine isolate of dentinogenesis imperfecta) in a 2½-year-old female patient from Prince Georges County, Md. The dental features bore a striking resemblance to trichodental syndrome, although the hair growth pattern and texture were not discussed by the authors. No genetic testing was performed, either. Hence, it is not certain if the cases "portrayed" as shell teeth in the past have anything in common with trichodental syndrome—other than the teeth's radiographic appearance.

Summary

Pediatric dentists play an important role in the identification and management of trichodental dysplasia patients. Both primary and permanent teeth may be affected by the condition. Consequently, the patient's family was alerted to expect abnormal permanent teeth after noticing the evidence from the patient's radiographs. Placement of stainless steel crowns (SSCs) on the permanent molars after eruption might be an option as well as potential extractions of permanent teeth—should they become symptomatic—as endodontic therapy is not an option. Although SSCs can protect the fragile shell teeth from possible caries or periodontal disease and help maintain the patient's vertical dimension, they can potentially interfere with the eruption of permanent teeth by prolonging the normal exfoliation of primary teeth. If the permanent teeth are lost, root form implants and/or dentures can be considered. The family was advised to have genetic counseling due to the syndrome's hereditary nature. This is because, for most autosomal dominant genetic conditions, there is a 50-50 chance for each child born to an affected individual to inherit the condition—regardless of the parent's or child's sex. Prenatal diagnosis for this syndrome is not yet possible. The patient is currently on a 6-month recall.

Acknowledgement

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Abstract of the Scientific Literature

Developmental Disturbances in Permanent Incisors After Deciduous Tooth Avulsion

Avulsions have been reported to make up 7% to 13% of all traumatic injuries in the deciduous dentition. This study aims to evaluate the frequency and type of developmental disturbance present in permanent incisors after traumatic avulsion of the respective primary tooth. Dental records of 4,238 children from the Danish Municipal Dental Health Service provided data on 35 children (0.8% of the study population) with 44 avulsed teeth. Only 30 of 44 permanent successors were fully erupted and, consequently, evaluated. These 30 permanent incisors were examined for developmental disturbances classified as: (1) discoloration; (2) hypoplasia; (3) horizontal linear hypoplasia; or (4) cervical dilacerations. Thirty percent of the studied teeth were found to have developmental defects, including 100% with discoloration, 9% with additional hypoplasia, and 6% with concurrent horizontal linear hypoplasia. Also, this study found that traumatic injuries occurring at younger ages were more closely associated with developmental disturbances in the permanent successor.

Comments: Parents need to be informed that developmental disturbances, including hypoplasia and discoloration, commonly occur in permanent incisors secondary to traumatic avulsion of the preceding primary tooth. **JMK**

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Christophersen P, Freund M, Harild L. Avulsion of primary teeth and sequelae on the permanent successors. Dent Traumatol 2005;21:320-323.

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