Trichodentoosseous (TDO) syndrome: case report and literature review

W. Kim Seow, BDS, MDSc, DDSc, PhD

Abstract

Trichodentoosseous (TDO) syndrome is an autosomal dominant condition characterized by dysplastic nails, curly hair, bone sclerosis of the long bones and calvarium, taurodontism, and enamel hypoplasia that occurs with hypomatureation/hypocalcification defects. To date, nine previous case reports of TDO in the dental literature have shown that while taurodontism and enamel defects are consistently present in TDO, the changes in the nails, hair, and bones may be variably expressed and occasionally not noted. Because of this, a few previous cases of TDO syndrome had been diagnosed simply as amelogenesis imperfecta. This case report highlights the typical clinical findings, diagnostic problems, and the clues to diagnosing this interesting condition. Clinical management of TDO is centered on preventing dental abscesses with stainless steel crowns and improving appearance with esthetic restorations. (Pediatr Dent 15:355–61, 1993)

Introduction and literature review

The term “trichodentoosseous” (TDO) syndrome first was coined by Lichtenstein et al. in 1972 to describe a large kindred with an autosomal dominant syndrome of curly hair, enamel hypoplasia and taurodontism, dysplastic nails, and generalized osseous dysplasia involving the long bones and calvarium. However, an earlier report in 1966 by Robinson and coworkers probably described the first case of the condition in the literature. To date, there have been only a few more cases reported.

TDO syndrome is of interest to dentists not only because of the severe enamel defects, but also because this condition is often confused with that of primary amelogenesis imperfecta. The reasons for the confusion may be that TDO syndrome is still not a well-recognized syndrome and that the other signs of this condition, i.e., bone, hair, and nail defects, are not always expressed, or may be minimal and often missed. Furthermore, the criteria for definitive diagnosis of taurodontism are not well established. These diagnostic problems have led to a few cases being published as amelogenesis imperfecta with taurodontism. The enamel is estimated to be approximately 1/4 to 1/8 normal thickness. These defects were associated with severe attrition, which may be why dental abscesses commonly are reported.

Histological examinations of affected teeth in TDO syndrome have shown the enamel to be undercalcified and thin. A scanning electron microscopic study reported the presence of pits and depressions on the enamel surface of the teeth. Furthermore, the pulp chamber may be enlarged, with the pulp horns extending close to the dentinoenamel junction, a condition similar to that seen in vitamin D-resistant rickets. Also, small amounts of interglobular dentin have been noted in a few teeth.

Taurodontism, a developmental condition in which the body of a tooth is elongated at the expense of the root (leading to a large pulp chamber) is consistently reported in all previous cases of TDO. A recent investigation, Seow pointed out that the taurodontism is always severe and involves all the molars, including the mandibular first permanent molar. An objective measurement of taurodontism in the mandibular first permanent molar could thus accurately diagnose TDO and distinguish it from the milder forms of taurodontism that may occur coincidentally in other types of inherited enamel defects.

Mode of inheritance

All previously published cases have shown that the defect is most likely transmitted in an autosomal dominant manner (Table).

Craniofacial defects

A few authors have reported frontal bossing as well as square jaw, mandibular prognathism, and dolichocephaly, as part of the craniofacial defects observed in TDO syndrome. However, the lack of cephalometric data makes it difficult to determine if these can be considered consistent findings in the TDO syndrome.

Dental defects

The dental defects in TDO are usually the most dramatic and consistent of all the signs (Table). Previous authors reported the teeth to be yellow-brown in color, displaying hypocalcification/hypomatureation enamel defects together with enamel hypoplasia. The enamel is estimated to be approximately 1/4 to 1/8 normal thickness. These defects were associated with severe attrition, which may be why dental abscesses commonly are reported.

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Table. Characteristics of families with the trichodentoosseous syndrome

<table>
<thead>
<tr>
<th>Author/Year</th>
<th>Inheritance</th>
<th>Frontal Bossing</th>
<th>Jaw Changes</th>
<th>Dental Abscesses</th>
<th>Dental Attrition</th>
<th>Teeth Color</th>
<th>Enamel Defects</th>
<th>Taurodontism</th>
<th>Hair Defects</th>
<th>Nail Defects</th>
<th>Bone Sclerosis</th>
<th>Other Changes</th>
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<tbody>
<tr>
<td>Reported as TDO</td>
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<tr>
<td>Robinson et al. (1966)²</td>
<td>AD</td>
<td>NR</td>
<td>Mand.</td>
<td>P</td>
<td>P</td>
<td>Yellow-brown</td>
<td>P</td>
<td>P</td>
<td>Curly, dry</td>
<td>P</td>
<td>A</td>
<td>NR</td>
</tr>
<tr>
<td>Lichtenstein &amp; Warson (1971)³</td>
<td>AD</td>
<td>NR</td>
<td>NR</td>
<td>P</td>
<td>NR</td>
<td>Brown</td>
<td>P</td>
<td>P</td>
<td>Kinky</td>
<td>P</td>
<td>P</td>
<td>Skin lesions</td>
</tr>
<tr>
<td>Lichtenstein et al. (1972)⁴</td>
<td>AD</td>
<td>P</td>
<td>Dolichocephaly, square jaw</td>
<td>P</td>
<td>P</td>
<td>Brown</td>
<td>P</td>
<td>P</td>
<td>Kinky</td>
<td>P</td>
<td>P</td>
<td>Clinodactyly</td>
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<tr>
<td>Jorgenson and Warson (1973)⁵</td>
<td>AD</td>
<td>NR</td>
<td>NR</td>
<td>P</td>
<td>NR</td>
<td>Yellow</td>
<td>P</td>
<td>P</td>
<td>NR</td>
<td>NR</td>
<td>NR</td>
<td>Impacted teeth</td>
</tr>
<tr>
<td>Gulmen et al. (1976)⁶</td>
<td>AD</td>
<td>NR</td>
<td>NR</td>
<td>P</td>
<td>NR</td>
<td>Brown</td>
<td>P</td>
<td>P</td>
<td>Curly</td>
<td>NR</td>
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<tr>
<td>Melnick et al. (1977)⁷</td>
<td>AD</td>
<td>NR</td>
<td>NR</td>
<td>NR</td>
<td>NR</td>
<td>Yellow-brown</td>
<td>P</td>
<td>P</td>
<td>Curly*</td>
<td>NR</td>
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<td>NR</td>
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<tr>
<td>Shapiro et al. (1983)⁸</td>
<td>AD</td>
<td>P</td>
<td>NR</td>
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<td>NR</td>
<td>NR</td>
<td>P</td>
<td>P</td>
<td>Wavy</td>
<td>P</td>
<td>P</td>
<td>Impacted teeth</td>
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<tr>
<td>Qualtromani et al. (1983)⁹</td>
<td>AD</td>
<td>NR</td>
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<td>P</td>
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<td>P</td>
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<td>Curly</td>
<td>P</td>
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<td>NR</td>
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<tr>
<td>Ogden (1988)⁹</td>
<td>AD</td>
<td>NR</td>
<td>Dolichocephaly</td>
<td>NR</td>
<td>NR</td>
<td>Yellow-brown</td>
<td>P</td>
<td>P</td>
<td>A</td>
<td>A</td>
<td>A</td>
<td>NR</td>
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<tr>
<td>Seow (present case)</td>
<td>AD</td>
<td>P</td>
<td>A</td>
<td>P</td>
<td>Yellow-brown</td>
<td>P</td>
<td>P</td>
<td>Curly</td>
<td>P</td>
<td>P</td>
<td>P</td>
<td>NR</td>
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<tr>
<td>Likely to be TDO¹ but reported as amelogenesis imperfecta</td>
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<td>Crawford (1970)¹³</td>
<td>AD</td>
<td>NR</td>
<td>NR</td>
<td>NR</td>
<td>P</td>
<td>B-Yellow</td>
<td>P</td>
<td>P</td>
<td>Kinky*</td>
<td>NR</td>
<td>NR</td>
<td>NR</td>
</tr>
<tr>
<td>Parker et al. (1975)¹⁴</td>
<td>AD</td>
<td>NR</td>
<td>NR</td>
<td>P</td>
<td>P</td>
<td>B-Yellow</td>
<td>P</td>
<td>P</td>
<td>NR</td>
<td>NR</td>
<td>NR</td>
<td>P¹</td>
</tr>
<tr>
<td>Congleton &amp; Burkes (1979)¹⁵</td>
<td>AD</td>
<td>A</td>
<td>A</td>
<td>NR</td>
<td>NR</td>
<td>Yellow</td>
<td>P</td>
<td>P</td>
<td>Wavy</td>
<td>A</td>
<td>A</td>
<td>NR</td>
</tr>
<tr>
<td>Elzay and Chamberlain (1986)¹⁶</td>
<td>AD</td>
<td>A</td>
<td>A</td>
<td>P</td>
<td>NR</td>
<td>P</td>
<td>P</td>
<td>Kinky</td>
<td>A</td>
<td>NR</td>
<td>NR</td>
<td></td>
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</tbody>
</table>

P — Reported as present; A — Reported as absent; NR — Not reported; * Curly at birth, straight at time of examination; † As reported by Witkop (1989)¹⁰; ‡ As suggested by Seow (1992).¹²
Nail defects

Nail defects include splitting of the superficial layers of the nails.\textsuperscript{1,3,7-8} In an affected individual, only some toenails may be affected.\textsuperscript{19} There is also variability in the expression of nail defects among TDO patients (Table).

Hair defects

Kinky or tightly curled hair may be a characteristic feature in many families\textsuperscript{1,3,5,8,13,16} (Table) and may be a distinguishing feature in diagnosing TDO from hypomaturation type amelogenesis.\textsuperscript{10,13,16} Other families have been reported to have wavy hair\textsuperscript{7,15} or curly hair at birth that straightened out later.\textsuperscript{9} Like the nail defects, the hair changes may vary considerably among affected members of the same family.

Bone changes

Sclerosis of the cortical bone had been noted in five of eight families with TDO,\textsuperscript{1,3,7,8,14} reported to be absent in three families,\textsuperscript{2,6} and not mentioned in another five families\textsuperscript{4,5,9,13,16} (Table). Bone sclerosis also may be a variable feature among members of an affected family.\textsuperscript{1,3} The most commonly reported areas of bone sclerosis include the skull base, mastoids, and zones of provisional calcification in the long bones.\textsuperscript{19} Bone thickening may lead to macrocephaly\textsuperscript{7} and has been associated with fractures.\textsuperscript{19}

Other changes

Other abnormalities that had been reported in previous TDO cases include impacted teeth,\textsuperscript{4,7} clinodactyly,\textsuperscript{1} and skin lesions.\textsuperscript{2}

Case report

Medical history

The previously undiagnosed propositus, a Caucasian female, was 9 years old when she was referred to the author by her general dental practitioner for management of discolored teeth. She was the product of an uncomplicated, full-term pregnancy resulting from a nonconsanguineous marriage. Her health history included only mild asthma for which she was treated with salbutamol and topical steroid aerosol sprays.

Family history

The patient appeared to have inherited the condition from her mother, who had similarly affected teeth (Fig 1). Three of the mother's brothers (not examined) were apparently unaffected. The mother was unsure as to whether the child's maternal grandmother was affected because she had lost contact with her. The maternal grandfather was apparently unaffected.

General features

On presentation at 9 years old, the patient appeared well nourished, weighed 28.5 kg (50th percentile) and was 132.0 cm tall (50th percentile). She had dry, curly hair (Fig 2a), which, according to the history, had become more curly in the previous two years. Her forehead was broad and appeared prominent on lateral view (Fig 2b). The mandible appeared slightly prognathic. Examination of the fingers and toes showed splitting of the free borders of the nails of several fingers and toes (Fig 3).

Skull and long-bone radiographic examinations revealed no bone abnormalities except for a slightly wider lateral skull diameter. A lateral skull radiograph showed a mild deficiency in midface development and a mildly prognathic mandible. A hand-wrist radiograph suggested normal bone age.

Dental features

Dental examination revealed partial eruption of all the permanent first molars, maxillary permanent central incisors, and mandibular permanent central and lateral incisors. The primary teeth present were the maxillary primary canines and left first molar. All the other primary teeth had exfoliated naturally or been extracted due to dental abscesses.

The permanent teeth were opaque, yellow-brown, and appeared poorly calcified. Enamel hypoplasia was very severe, particularly in the permanent first molars (Figs 4a, 4b). In contrast, the remaining maxillary primary canines and left first molar appeared less affected, although there...
was severe attrition of the primary maxillary left first molar.

Oral hygiene was poor with gross amounts of plaque and associated gingivitis. Both posterior and anterior occlusal relationships revealed a Class III tendency.

At the age of 12 years, additional photographs were available, showing all permanent teeth to be similarly affected by the hypomaturation/hypocalcified-hypoplastic type of enamel defects (Figs 5a, 5b, 5c).

**Dental radiographic findings**

An orthopantograph exposed at age 12 years (Fig 6) showed very mild sclerosis of the mandibular cortical bone and very mild increase in bony trabeculation. All the permanent teeth, including third molars, were present. The first and second molar teeth appeared to be taurodont.

Furthermore, an objective assessment of taurodontism in the mandibular first permanent right and left molars yielded crown/body-root length ratios of 1.30 and 1.55 respectively, thus confirming the presence of meso-taurodontism. In addition, the pulp chambers were large, mostly due to a marked decrease in the thickness of dentin. The enamel appeared extremely thin or even nonexistent in some teeth, with little radiographic contrast between enamel and dentin.

**Examination of the mother**

The mother of the propositus was a single parent aged 35 years.
31 years at the time of dental examination. She had wavy hair since youth, and all her fingernails and toenails were thin and split at the free borders. She presented with a dolichocephalic face, and a mildly prognathic mandible (Figs 7a, 7b). A skull radiograph revealed mild sclerosis of the vault and base of the skull (Fig 8).

On dental examination, only the mandibular incisors, canines, first premolars, left second premolar, and right third molar were present. The other teeth apparently had been extracted at a young age due to dental abscesses. Like her daughter's dentition, the teeth were yellow-brown, and showed typical hypomaturation/hypocalcification enamel changes together with enamel hypoplasia (Fig 9). Moderate gingival recession was observed. A full maxillary denture was worn.

**Dental treatment**

The patient was treated at the graduate students' clinic for restorative dental work.

At the time of dental examination, the patient complained of severe thermal sensitivity of the molars. Stainless steel crowns were cemented with glass ionomer cements after minimal preparation. At age 11 years when the second premolars were beginning to erupt, maxillary removable appliances were used successfully to distalize the maxillary first permanent molars to regain spaces lost through premature loss of the maxillary primary molars. At age 12 years, the stainless steel crowns on the first molars were replaced and crowns placed on the second molars. The hypoplastic and carious defects on the distal surfaces of the maxillary first premolars were restored with glass ionomer silver restorations. At that time, it was planned to use acid-etched composite veneer restorations to improve the appearance of the anterior teeth as soon as the minor orthodontic corrections were completed. The patient was not particularly concerned about the appearance of her anterior teeth at that time.

**Discussion**

This report of a young patient and her affected mother highlights many important features of the trichodentoosseous syndrome. The curly hair and nail defects were clearly evident. The teeth were typically taurodont to a severe degree, and the enamel defects were of the hypomaturation-hypocalcification type with enamel hypoplasia. The bone defects, however, were expressed variably. Mild bone sclerosis was manifested at the base of the skull in the mother, but in the daughter was observed only to a very mild degree in the mandible. These hair, nail, and dental defects were similar to those reported previously by various authors (Table).

This report, as well as those cases studied previously, show clearly that dental defects constitute the most significant signs of the TDO syndrome. Yellow-brown, opaque discoloration of the enamel—resulting most likely from defects in hypomaturation/hypocalcification—appear to be the most consistent feature in all cases reported. The enamel is also thin and easily abraded. Although one study had indicated the occasional presence of interglobular dentin, in all other reports that included histological examinations, as well as this case, there were no structural changes in dentin. This fact is of interest since syndromes involving bone defects are usually accompanied by changes in dentin. In this case, however, the dentin thickness is reduced, thus resulting in very large pulp chambers, easily exposed, because the soft enamel is prone to abrasion.

Taurodontism is another feature consistently reported in TDO (Table). This developmental aberration may be observed in approximately 3–8% of the general population, but may be an accompanying sign in dental hypodontia and ectodermal dysplasia, Down, Klinefelter, and other dysmorphic syndromes. The degree of taurodontism in TDO has been noted to be great and Seow recently proposed that the severity of taurodontism—as well as the involvement of the man-
dibular first permanent molars—may be used to accurately diagnose TDO syndrome.

A few cases of amelogenesis imperfecta of the hypomaturation type occurring with severe taurodontism have been reported in the dental literature.\textsuperscript{5,6,10,11} Of these, three\textsuperscript{6,10,11} have since been diagnosed as TDO by Witkop,\textsuperscript{12} based on the history of hair and/or nail defects. The fourth family, reported by Congleton and Burkes\textsuperscript{15} also recently has been suggested to be TDO,\textsuperscript{12} based on accurate diagnosis of taurodontism using an objective measurement technique, as well as the inclusion of wavy hair in the family as an additional feature not previously considered by other authors.

Diagnostic criteria

Diagnosis of TDO is not always clear cut, and to aid diagnosis it is proposed that a positive identification of TDO should include the main criteria: 1) generalized enamel defects that show hypomaturation or hypocalcification occurring with enamel hypoplasia; 2) severe taurodontism of the teeth, involving the mandibular first permanent molars; 3) an autosomal dominant mode of inheritance and at least one of the other features (i.e., nail defects, bone sclerosis, and curly, kinky, or wavy hair present at a young age, that may straighten out later).

Management strategies

The main clinical problems experienced by patients with TDO syndrome are poor dental esthetics, sensitivity of the teeth, loss of occlusal vertical dimension from loss of tooth structure, and dental abscesses from pulp exposures. The main aim in managing affected patients centers on preventing clinical problems as early as possible and improving dental esthetics.

The discoloration of the teeth may need full-coverage jacket porcelain crowns. However, this type of restoration may not be recommended in the young patient due to large pulps. Interim veneer restorations using bonded composite resins or porcelain may be considered. In the present case, the decision was made to wait until completion of minor tooth movement before anterior restorations since the patient was not particularly concerned about appearance. Sensitivity of teeth and excessive loss of tooth structure leading to pulp exposures and decrease of occlusal vertical dimension may be prevented by stainless steel crown coverage as soon as the molars have reached adequate crown height. The crowns may involve a conservative technique\textsuperscript{15} in which no crown reduction is performed. In this method, glass ionomer cement or composite resin is placed on the occlusal surface of a partially erupted molar so that the tooth erupts with the resin in occlusion with the opposing teeth. In the preparation of the steel crowns, occlusal reduction is then performed on these resins only. Proximal reduction on the molar teeth is obviated by the prior placement of separating elastics through the contact points. Glass ionomer cements are the choice for crown cementation. The steel crowns may be replaced later with permanent gold crowns.

Restoration of carious and hypoplastic defects is achieved with adhesive materials if possible since the weak enamel margins tend to fracture away from nonadhesive dental amalgams.

Previous cases indicate that many affected adults had early loss of teeth from extractions due to dental abscesses. Early diagnosis of the condition and early institution of prophylactic crowns would prevent these tooth losses.

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Trauma centers should take the lead in preventing violence against women

Violence against women and girls is an "unaddressed imperative and an indictment of the moral character of our society," according to a study published in the June issue of the AMA’s Archives of Surgery. Rosalyn P. Sterling-Scott, MD, Department of Surgery, Charles R. Drew University of Medicine and Science, Los Angeles, Calif., and colleagues, collected data on firearm fatalities against females in California from 1987 through 1991, and among those injured by firearms and treated at the Los Angeles County-Martin Luther King Jr./Charles R. Drew Trauma Center (KDMC) from September 1, 1991 through December 31, 1992.

The researchers found that among the female firearm deaths in the state of California between 1987 and 1991, there was a steady upward trend for all ethnic groups except black women. The study says: "Most of the fatalities were recorded for those aged 20 to 39 years. The overall relationship among the age groups seems to be relatively constant except for those aged 10 to 19 years. In this group, there has been a 2.8-fold increase in deaths from 1987 to 1991."

Of the 1,862 patients treated at the KDMC for firearm injuries, 153 (8.2%) were female. The girls and women ranged in age from one to 73 years, with a mean age of 27 years. Ninety-nine (65%) were black and 54 (35%) were Hispanic.

Thirty-two patients (21%) required at least one major procedure. Of the 16 patients who died, 14 (88%) died in the emergency department of wounds to the head (six patients), chest (five patients), and abdomen (three patients). The researchers write: "The high prevalence of violence against girls and women of all ages brings them into regular contact with physicians. Unfortunately, physicians tend to treat the injuries symptomatically and rarely probe for the underlying causes. "They point out that much of the violence against girls and women is perpetrated by those known to them."

They say: "Trauma centers must take the lead in the collection, organization, computerization, and analysis of meaningful data surrounding violent injury." They add: "In addition to gathering data, trauma centers offer important opportunities for the initiation of interventions at the time of care. Since we know that the likelihood is great for a woman to be intentionally injured by someone she knows or with whom she has been intimate, screening for current and past victimization should be routine."

The authors call upon trauma surgeons to "become visionaries who transform their trauma centers into centers of healing and intervention."