Osteogenesis imperfecta with dentinogenesis imperfecta: a mistaken case of child abuse

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Abstract

Mandatory reporting of child abuse became a state law throughout the United States during the 1970s causing a great increase in the number of cases investigated annually. Before confirming a diagnosis of child abuse, conditions which present signs similar to abuse should be considered and investigated. Unexplained bone fractures, for example, often seen in child abuse, also may be the initial presenting feature of osteogenesis imperfecta (OI). A case of suspected child abuse is presented which evaluation revealed to be OI. The oral examination provided the first documented evidence, other than fractures, that OI existed. Dental evaluation revealed several features which were consistent with dentinogenesis imperfecta (DI). The teeth had a yellow-brown discoloration which appeared opalescent in the cervical area. Large pulp chambers with no obliteration and only slight crown belling were seen in the posterior teeth. Extensive wear and enamel fracturing were prevalent in the anterior teeth. In cases of unexplained skeletal fractures or suspected OI it is recommended that a thorough dental evaluation be performed to confirm the presence or absence of diagnostic oral features.

Child abuse only recently has become a public responsibility -- in the past it was considered a private issue. Mandated reporting of child abuse was enacted in the United States during the 1970s. New reporting policies, growing awareness in the medical community, and establishment of child protection agencies greatly increased the number of cases investigated annually.

The most common forms of child abuse are deprivation of necessities and emotional maltreatment; major physical injury and sexual maltreatment make up a small portion of the cases reported. Child abuse associated with physical injury, often referred to as the battered child syndrome, usually is manifest in the form of contusions, abrasions, lacerations, burns, dental injuries, dislocations, and fractures.

Many cases of child abuse will appear characteristic while others may be extremely difficult to diagnose. For this reason strict reporting practices and multidisciplinary teams have been established to manage child abuse and neglect. Before a diagnosis is confirmed, an evaluation of conditions which may mimic child abuse should be considered. A differential diagnosis may include metabolic abnormalities, hematologic disorders, skeletal pathologies, and accidental trauma. The parents and the child may be damaged socially and psychologically as a result of misdiagnosing a case as child abuse.

Dental evaluation is indicated in cases where facial trauma exists or in pathologic conditions that present diagnostic features in the oral cavity. A case of suspected abuse in a child who actually had osteogenesis imperfecta (OI) will be presented and the importance of the dental examination discussed.

OI represents a group of inherited disorders characterized by bone fragility, blue sclera, deafness, dentinogenesis imperfecta (DI), skin and vessel fragility, and hyperelasticity of the ligaments. Considerable controversy has existed concerning OI's mode of transmission. Early reports by Apert and Fuss showed that the disease was transmitted as an autosomal dominant trait according to Mendelian inheritance. Some authors considered OI to be inherited through a single pleiotropic gene with variable penetrance and expressivity which ranged from simple osteoporosis to the triad of blue sclera, bone fragility, and deafness. Sporadic cases were attributed to new spontaneous mutations. Recent research by Sillence has resulted in a classification system of OI based on clinical expression and inheritance patterns. He divided OI into four basic types, two autosomal dominant and two autosomal recessive.

DI may occur as an independent single autosomal dominant defect of the dentition or in conjunction with OI. Witkop suggests that the term hereditary opalescent
dentin be used when the disease is seen as an isolated trait, and if found with OI, dentinogenesis imperfecta should be used.® Shields and Bixler have classified DI associated with OI as Type I and the dental anomaly alone as DI Type II.®

Due to the variable clinical appearance of the dentition and diverse classification systems for OI it is difficult to assess the frequency of DI occurring with OI. The classic features of DI include bell-shaped crowns, short blunt roots, an opalescent blue gray enamel hue, pulp obliteration, and rapid wear as a result of enamel fracturing. The teeth, however, may range from being nearly normal in color, to yellow-brown, to the characteristic opalescent blue gray.® Pulp morphology also varies from being obliterated, to normal, to enlarged.®

Clinical Report

The male child presented in this report was the product of a full-term pregnancy and Caesarean section delivery. He was the only offspring born to a Caucasian couple with no history of consanguinity. The father had two children from a previous marriage, neither of whom were affected with OI or DI. No miscarriages or terminated pregnancies were reported by the mother.

The child’s postnatal history was uneventful until 19 months of age when he fell from a porch, resulting in a head injury and hospital admission. A left parietal hematoma and fracture of the premastoid complex was diagnosed which required surgical intervention. During this admission several healing rib fractures with callus formation were noted on a routine chest radiograph. The possibility of multiple traumas having unknown etiology was noted but not investigated.

Two days after discharge he was readmitted with a fracture of the right femur after falling at home. Radiographs of the long bones were obtained due to the possibility of child abuse and to rule out pathology. They showed a normal trabecular pattern and no evidence of epiphyseal injury which might indicate pathology. The child protection agency was contacted, abuse charges were filed due to multiple unexplained fractures, and the child was taken into protective custody by the state. After three months of court hearings, counseling sessions, and psychological testing (which indicated the parents were not likely candidates for child abuse) the child was returned to the mother and father.

At three years of age, and only 10 months after the parents regained custody, the child again was brought to the hospital. According to the parents the child fell to the floor while playing, apparently injuring his left leg. Physical examination revealed a slight swelling just above the ankle with no signs of bruising. Radiographic evaluation showed a fracture of the left tibia. After reviewing the patient’s past medical history, the case again was referred to social services which culminated in child abuse charges being filed against the parents. The child was returned to protective custody by the state.

Interviews with the family pediatrician at this time revealed no findings consistent with child abuse other than the fractures. Discoloration of the teeth had been noted by the pediatrician who suggested further evaluation for bone pathology. Radiographic follow-up of the long bones showed a generalized osteopenia which was not considered remarkable. A dental consultation was requested to evaluate the discolored teeth.

At the time of the initial dental examination several salient physical findings were revealed including marked distention of the lower rib cage, slight frontal bossing of the cranium, and a prominent occiput. Oral evaluation disclosed a full complement of primary teeth having a yellow-brown to gray hue (Figure 1). The incisors showed extensive attrition. Radiographs of the teeth revealed slightly bell-shaped crowns and wide pulp chambers without evidence of obliteration except in the mandibular incisors (Figure 2). In light of the dental, radiologic, and phenotypic findings, a written deposition was given to the social worker indicating OI was a possible diagnosis. On the basis of this testimony litigation against the parents was discontinued.

A referral was arranged for an interdisciplinary and genetic evaluation. Several new findings consistent with OI were elucidated at this time including hyperelasticity of the joints and slight kyphoscoliosis. A marked hearing loss was noted to the left ear (this resulted from the head trauma — not otosclerosis associated with OI). Due to the mildness of the case and the lack of OI in the family

![Figure 1. The primary dentition shows generalized discoloration and excessive wear on the anterior teeth.](image1)

![Figure 2. A radiograph of the teeth at four years of age shows slightly bell-shaped posterior crowns with large pulp chambers.](image2)
history, a new autosomal dominant mutation was felt to be the most likely mode of inheritance.

Discussion

Mild forms of OI which are difficult to diagnose may be mistaken for child abuse due to repeated episodes of unexplainable bone fractures. Paterson conducted a survey of 170 OI patients and found that nearly 6% had been cases of suspected child abuse. The genetic heterogeneity and clinical variability of OI often produce an elusive picture of signs and symptoms. The dental manifestations of this disease can provide significant information as illustrated by this case.

When trying to rule out or confirm OI associated with child abuse, a careful gathering of data through an interdisciplinary approach is necessary. In addition to the child protection agency (which must be contacted), a physician, orthopedist, geneticist, dentist, and psychologist should be consulted to aid in the diagnosis. In this case, it was not until the dental evaluation that substantial evidence implicating OI emerged.

Radiographic surveys may reveal undiagnosed fractures in various stages of healing as seen in this patient. The long bones are most commonly involved, but rib and skull fractures as well as spine deformities may occur. In mild OI cases the bones may appear normal radiographically, adding to the difficulty of reaching a diagnosis.

As in this case, not all DI associated with OI will present the classic manifestations of pulpal obliteration, opalescent discoloration, bell-shaped crowns, short roots, and rapid wear. The color changes may be subtle and appear more yellow brown than the characteristic blue gray. The pulp chambers, which are usually obliterated, may appear normal to enlarged as was seen in this patient. Pulp obliteration may not occur until later in life, so pulp chambers in young patients often will appear normal. The dentist must examine potential OI cases carefully so as not to overlook the variable expressions of DI Type I.

Conclusion

A case of suspected child abuse has been reviewed in which the dental evaluation provided information leading to a diagnosis of OI. It is recommended that a thorough dental examination be performed in cases of unexplainable fractures or suspected OI. The unfortunate mislabeling of cases as child abuse may be avoided through an interdisciplinary approach.

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References