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REVIEW ARTICLE

Deborah S. Ablin
Shashikant M. Sane

Non-accidental injury: confusion with temporary brittle bone disease and mild osteogenesis imperfecta

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D. S. Ablin
Department of Radiology,
University of California Davis Medical Center, 2516 Stockton Boulevard,
Ticon II, Sacramento, CA 95817, USA

S. M. Sane
Department of Radiology,
Children's Health Care Minneapolis,
Minneapolis, Minnesota, USA

Abstract Accurate diagnosis of non-accidental injury (NAI) can be reached in the majority of cases by careful appraisal of the social and family history, combined with painstaking clinical roentgenographic and other imaging evaluations. Careful review of the scientific literature clearly indicates that collagen analysis to exclude mild forms of osteogenesis imperfecta, especially type IV, is recommended only in rare cases in which diagnosis of NAI remains in doubt even after thorough evaluation by experienced radiologists and/or other physicians. Until clinical research scientifically establishes the existence of temporary brittle bone disease, it should remain strictly a hypothetical entity and not an acceptable medical diagnosis.

Introduction

When an infant or a child presents with one or more unexplained fractures, it is of crucial importance to determine whether the injuries are due to child abuse (non-accidental injury, NAI), or result from an underlying bone disease. To send a child home to the same abusive environment may result in his death or severe morbidity. To take a child away from his loving parents or guardians unnecessarily, when the child has an underlying bone disease, is a tragedy both for the child and the caregivers. Fortunately, in the majority of instances, the correct diagnosis can be reached by careful appraisal of social and family history and careful clinical and roentgenographic examination [1–4]. The entities confused with NAI are mild forms of osteogenesis imperfecta (OI), especially OI type IV, and a hypothetical variant of OI, namely temporary brittle bone disease (TBBD) [5].

Proposed temporary brittle bone disease

The so-called entity of TBBD, proposed as a variant form of OI, originated as a presentation at the Fourth International Conference of OI in 1990. An article was subsequently published in the *American Journal of Medical Genetics* without peer review [6]. Paterson et al. described 39 patients seen over a 10-year period with fractures occurring only in the first year of life. The authors speculated that "this disorder reflects a temporary collagen defect and is probably caused by a temporary deficiency of an enzyme, perhaps a metalloenzyme, involved in the post-translational processing of collagen" [5]. None of these three postulates in this single sentence have been substantiated by the authors with sound scientific data or subsequently corroborated in any peer-reviewed journals by the same or other authors with prospective scientific research data. In addition, the authors proposed cuproenzyme lysyl oxidase as a deficient enzyme in the disorder – once again, without any supporting credible scientific evidence. Copper deficiency was proposed as another explanation for TBBD. However, the serum copper level was measured in only three of the 39 patients with TBBD, and of these, two were normal [5].

Recent comments in an English court by Paterson cast grave doubt as to the methodology by which he included some cases in his series [7]. When specifically asked by Justice Wall how two earlier cases, ruled by the court as NAI, would be treated in his research data, Paterson an-

swered by saying both would be included in his research as proven cases of brittle bone disease [7–9]. When asked how the case being heard by Justice Wall would be recorded in his research if the court ruled NAI, he responded that the case would still be logged in his research data as TBBD [7]. The case – an infant with shearing contusional brain injury and multiple fractures of different ages which ceased in a protected environment, including metaphyseal corner fractures and posterior rib fractures – was subsequently ruled by the court as unequivocal NAI and inconsistent with any other diagnosis [7]. Paterson's statements in court cast the gravest doubt on the accuracy of his published research on TBBD [7, 8].

If abused children were actually included among those reported as having TBBD, this may explain why the data have characteristics typical of non-accidental trauma, such as rib and metaphyseal fractures observed in the first year of life. Periosteal reactions (especially symmetrical), expanded osteochondral junctions, and palpable liver, reported as characteristics of TBBD, are frequently seen in young infants as normal findings. Apnea, vomiting, and diarrhea in the first year of life, symptoms attributed to TBBD, are nonspecific and occur commonly. Furthermore, apnea, vomiting, and enlarged anterior fontanelle, features attributed to TBBD, may be warning signs of severe head injury in abused children [5, 7, 10]. Fractures occurring in the hospital, as described in seven cases of so-called TBBD, are a well-recognized phenomenon; experienced radiologists know that occult fractures, particularly those involving the ribs, may first become evident in the hospital as the callus ossifies [11]. No comprehensive detailed clinical information, detailed specific radiological findings of skeletal surveys, or other diagnostic imaging studies, other than general descriptions and three radiographs, are provided for the other 32 infants with TBBD, who were incidentally discovered to have fractures outside of the hospital. Hence, objective analysis of the data by an independent observer is not possible.

Osteogenesis imperfecta type IV

Type IV OI is autosomal dominant and is present in 5 % of patients with OI [12]. It embraces a heterogeneous group of patients who have OI without blue sclerae and who exhibit mild to moderately severe bone fragility. The majority of patients have a positive family history [12]. Although new genetic mutations may result in this type of OI and have no family history, such patients will frequently have wormian bones, osteoporosis, bowing deformity, thin cortices, short stature, ligamentous laxity, deafness, or dentinogenesis imperfecta [2]. Taitz [13] calculates the hypothetical incidence of OI type IV without family history or other features of the disease to be 1:1 000 000 to 1:3 000 000 live births. In a population of 500 000 with 6000 live births a year, the incidence of such a rare instance of OI type IV would be one case in 100–300 years. In such selected difficult cases, skin biopsy for collagen analysis from cultured dermal fibroblasts may help to identify children with OI [14, 15]. Unfortunately, this test is not definitive. The false-negative rate for patients with all types of OI is 13.6 %, although it is lower (4.5 %) for OI type IV, as reported by Wenstrup et al. [14]. Furthermore, the presence of OI does not exclude the possibility of child abuse as the cause for fractures. Simultaneous child abuse and OI have been reported [16, 17].

Steiner et al. reviewed the collagen analysis in children referred for distinction of OI from child abuse [15]. They concluded that in the majority of cases collagen analysis was not necessary. Those children suspected as victims of abuse with abnormal collagen analysis and OI could usually be diagnosed by careful clinical evaluation by experienced radiologists/physicians. Those referred children with physical findings pathognomonic of abuse and not part of the spectrum of OI had normal collagen analysis. Collagen analysis was recommended only in those rare children in whom the diagnosis remained in doubt after careful clinical and roentgenographic evaluation [15].

Safety of the child paramount

The reports from the states to the National Center on Child Abuse and Neglect [18] published in July of 1996 indicate that the number of abused and neglected children in the USA rose sharply from 1.4 million in 1986 to 2.9 million in 1993. It is the responsibility of all child advocates to err on the side of safety and place the child in protective custody if the diagnosis of NAI is being seriously considered, while a rare or mild form of OI, such as type IV, is being excluded. Collagen analysis may take as long as 3 months. During a period of protection, significant changes on follow-up skeletal surveys may occur which leave the diagnosis of OI no longer in doubt. In many instances, unexplained fractures will cease to occur in a protected environment and observed osteopenia may gradually disappear with healing, indicating strongly the diagnosis of NAI, not OI or TBBD.

Conclusion

It is our view that until clinical research scientifically establishes the existence of TBBD, it should remain strictly a hypothetical entity and not an acceptable medical diagnosis.

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