underprivileged circumstances NAI is always assumed as the cause for inexplicable fractures; that the child with osteogenesis imperfecta is not immune from NAI (see Knight and Bennett[16]); that none of their three patients with osteogenesis imperfecta had osteopenia or wormian bones; and that a history incompatible with injury is as typical of osteogenesis imperfecta as NAI. They recommend fibroblast culture in infants or children who present with fractures that suggest NAI – first, if there are no external signs of child abuse (such as bruises or head injury); second if the fracture site is consistent with the history, but the mode of injury seems too minor to have caused a fracture; or third if the child has had fractures in different environments. Since the culture of fibroblasts from skin biopsy and subsequent collagen analysis can take at least three months it is important that (where appropriate) the infant is in a safe place during that time. Apart from providing protection this period is not merely spent in waiting for the collagen result, as important changes in the skeleton can occur so rapidly that the diagnosis of osteogenesis imperfecta is soon no longer in doubt.

(5) Court proceedings

Expert witnesses may wish to win their case, but this should not be done at the expense of the facts and is not necessarily in the best interests of the child. In cases of NAI the approach should be inquisitorial rather than adversarial.[17] In an ideal world the view given by the expert should be straightforward, not misleading or biased and well researched; a well balanced and non-partisan view will be more welcome to the court than fixed ideas and an inability to consider all sides of the problem.

(6) Conclusion

The distinction between NAI and osteogenesis imperfecta is a small and untidy corner of paediatrics. Osteogenesis imperfecta is rare and few people have extensive experience of it; the views of those who have should be taken into account, irrespective of their specialty. Where legally necessary biochemical confirmation of osteogenesis imperfecta should be sought. The suggestion that a form of temporary skeletal fragility is due to copper deficiency requires more investigation.

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Commentary (1)

Dr Smith has reopened the debate as to the differential diagnosis of bony disorder in infancy by looking at the advances made, particularly in the understanding of the underlying collagen disorder in osteogenesis imperfecta. Unfortunately the clinical investigation of type IV osteogenesis imperfecta has not been undertaken with similar scientific rigor and a description of this variant is required.[1]

Taitz wrote that if osteogenesis imperfecta occurs in 1/20 000 births and type IV osteogenesis imperfecta occurs in 5% of this total the occurrence of osteogenesis imperfecta with no family history or blue sclera would be in the order of 1/3 000 000 births.[2] One to two per cent of all children will be physically abused, although only 5–10% will sustain fractures.[3]

The diagnosis of physical abuse requires much more than an opinion of a radiograph or even a fibroblast culture. Abuse may occur in families with organic disorders, hence the requirement to carefully build up the diagnostic jigsaw (as Dr Smith says), but this must also be viewed in the wider social context too. There is also more to be understood in the relationship between the biochemical abnormality of the collagen, clinical disease, and fracture. Adults with collagen abnormality do not necessarily have a past history of fractures and current techniques of investigating collagen disorders are time consuming and offer a research rather than a clinical tool.

Fractures in 'mild' disease are, by definition, associated with weight bearing or trauma – rib, skull, and metaphyseal fractures do not occur spontaneously.[4] Where metaphyseal fractures have been reported in osteogenesis imperfecta it has been in the context of other skeletal abnormality.[5]

Although the skeleton may be apparently normal, wormian bones are common even in 'mild osteogenesis imperfecta', even if they are not apparent at birth or in the immediate postnatal period.

The question of temporary brittle bones

remains unproved. The series of Paterson *et al* contains too little clinical information – 39 patients over 10 years were diagnosed, seven infants sustained fractures in hospital (were these the 21% born at less than 33 weeks gestation?).[6] The fractures were numerous, rib fractures were common as were metaphyseal abnormalities and periosteal reactions. Three had fractures at birth, and most at less than 4 months. How the child came to have radiographs taken is not clear or how physical abuse was excluded. The picture has similarities to the copper or temporary copper deficiency story which was put into perspective by Shaw in 1988.[7]

Dr Smith concludes by acknowledging that osteogenesis imperfecta is rare. He might have added that osteogenesis imperfecta with no family history of fractures, joint laxity, early onset deafness, blue sclerae, and dentinogenesis is very rare. In addition the probability that an individual infant with no relevant family history has osteogenesis imperfecta where the skeleton is normal, there are no wormian bones, there is no or trivial history of trauma, the infant is not weight bearing yet has a fractured skull, ribs, or metaphyseal fractures is in the Taitz range of probabilities, that is, millions.

Collagen analysis is clearly important in understanding the pathogenesis of osteogenesis imperfecta, but until the correlation with clinical disorder is more precise it is difficult to see the biopsy report being more than another piece of the jigsaw, that is, not a diagnostic test but offering some corroboration of non-abuse. The diagnosis of possible osteogenesis imperfecta can not wait three months but a multicentre study looking at fractures in infancy (paediatrically, radiologically, orthopaedically, and biochemically) would be the way forward to sort out the real from the supposed disorder.

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Commentary (2)

This review has been invited to try and establish the parameters for making the diagnosis of osteogenesis imperfecta in order to avoid an erroneous diagnosis of NAI and to review the evidence for the most recently suggested hypothesis, temporary brittle bone disease,[1] put forward as an explanation in contested cases of NAI. Has the article achieved its objectives and how does it take the debate forward? I offer the following comments.

The level of general knowledge about current concepts in osteogenesis imperfecta is very varied within paediatrics, and developments in molecular genetics in establishing the presence of abnormal collagen in up to 85% of patients with osteogenesis imperfecta are perhaps not as widely appreciated as they should be. The Sillence classification remains the one most understood within paediatrics. Dr Smith indicates that not all patients fit comfortably within the mould. We still await a definitive article in the general paediatric literature, explaining new thoughts on osteogenesis imperfecta classification, broadly referenced from several sources. We also need to understand why 15–20% of patients with osteogenesis imperfecta do not have the gene for abnormal collagen. Is there another genetic problem as yet undiagnosed? Smith states that the normal population do not have abnormal collagen but the statement is unreferenced. A reference indicating the basis for this would be helpful. A greater general understanding of the genetics of osteogenesis imperfecta, and the place of other collagen disorders demonstrable by fibroblast culture in relation to osteogenesis imperfecta, is of immense importance. There are still many unanswered questions.

Dr Smith indicates that recent reviews indicate that type IV osteogenesis imperfecta, and I presume he refers to both types A and B, is more frequent than thought. That statement is unreferenced. We need to know the evidence. It would be extremely helpful to know the incidence and natural history of type IV A osteogenesis imperfecta, the form usually suggested in contested cases of abuse, quoting references other than Paterson and Taitz.[2–4] In Sillence's paper[4] he indicated then that the radiology of the skeleton in type IV has not been well defined, the bone changes were severe, and he comments that there is no known feature differentiating type IV from osteogenesis imperfecta type III. Since then, it has been suggested that the radiographic features are more variable.[1]

Paterson's diagnosis of type IV A osteogenesis imperfecta has been questioned on the basis of his methodology.[5-7] Therefore, we need independent sources. Taitz's calculation of the incidence of new mutations is the standard reference.[3] Is this accepted as accurate by geneticists, for, if so, then the possibility of a wrong diagnosis being made in contested cases becomes so unlikely that it hardly merits discussion?

Dr Smith indicates that no type of fracture excludes osteogenesis imperfecta and that infants with it may sustain any type of fracture in a skeleton that is radiologically otherwise normal. What is the evidence for this? Children with severe changes of osteogenesis imperfecta may have multiple fractures affecting almost anywhere, excepting the skull, unless there is coincidental trauma. Children with mild disease, on the contrary, mainly sustain