Brittle or battered

There has recently been considerable comment in the national and medical press about the differential diagnosis of the fractures of non-accidental injury in infants,\(^1\)\(^-\)\(^3\) and it has been stated that many of these infants are suffering from copper deficiency or a mild form of osteogenesis imperfecta. Clinical experience and a careful review of the literature, however, would indicate that these conditions should not be confused with non-accidental injury.

The diagnosis of non-accidental injury is not made lightly, but certain clinical findings must raise the question of the diagnosis. These include torn frenulum of the lip, unexplained bruising, retinal haemorrhages, and delay in seeking medical attention for fractures. In these circumstances radiography of the skeleton is part of the routine clinical assessment. The finding of metaphyseal fractures or fractures in different stages of repair in the presence of normal bones without a history of injury is regarded as being diagnostic of non-accidental injury. Other fractures that should strongly suggest non-accidental injury are comminuted wide skull fractures, scapular fractures, and fractures of the outer end of the clavicle. Similarly it is strongly suggested by rib fractures.\(^4\)\(^-\)\(^8\) Rib fractures only occur in children with normal bones in response to severe trauma such as road traffic accidents. Even the compression forces used in vigorous cardiopulmonary resuscitation rarely produce them. In one series of 113 children no rib fractures were found in children who had undergone cardiopulmonary resuscitation.\(^9\) I have never personally seen a fractured rib after this procedure.

A full review of the radiological manifestations of child abuse can be found in a recently published book by Kleinman.\(^10\) It has been said that the only reason a differential diagnosis is considered is the reluctance to accept the implications of the bony lesions.\(^11\)

Copper deficiency

A full review of copper deficiency is published elsewhere in this issue (p 448). This annotation gives a synopsis of the salient clinical and radiological features. The reported clinical features of copper deficiency in infants are psychomotor retardation, hypotonia, pallor, hypopigmentation of skin and hair, prominent scalp veins, sideroblastic anaemia resistant to iron treatment, and neutropenia (J C L Shaw, personal communication).\(^12\)\(^-\)\(^19\) The radiological changes are those of osteoporosis, fraying and cupping of the metaphyses, spur formation, periosteal reaction, and fractures.\(^12\)\(^-\)\(^26\)

About 40% of reported cases of copper deficiency are in premature infants of whom about 25% are on long term parenteral nutrition (J C L Shaw, personal communication).\(^12\)\(^-\)\(^19\) There have been cases in full term infants who have had a severe dietary deficiency of copper.\(^18\)\(^19\)\(^27\)\(^28\) The median age at presentation is a mean of 8-3 months for full term infants and 3-0 months for preterm infants. Copper deficiency has never been described in an infant fed exclusively breast milk or in a full term infant fed a formula known to contain adequate amounts of copper. Since 1983 most, though not all, formula milks freely available in the United Kingdom contain enough copper to prevent dietary deficiency.

Plasma copper concentrations are low at birth but reach adult concentrations by 4 months\(^29\); they can be reliably measured by atomic absorption spectrophotometry. When this method is used no child has been reported with the syndrome of copper deficiency with a plasma concentration of greater than 43 \(\mu\)g/dl for term infants and 33 \(\mu\)g/dl for preterm infants (J C L Shaw, personal communication). In all cases the response to treatment either by the introduction of copper supplements or dietary improvement has been prompt with a reticulocytosis, a rise in haemoglobin and neutrophil count, and resolution of the bone changes. There are no reported cases of fractures occurring as a late sequel to proven copper deficiency.

The bone disease of copper deficiency is a metabolic disease. The osteoporosis affects all bones and the metaphyseal fraying affects the growing ends.\(^12\)\(^-\)\(^25\) These features are most easily seen in the distal radius and ulna, though the fraying is more pronounced in the ulna.\(^22\) The fraying leads to sickle shaped spur formation and fractures may occur through these. These fractures do not resemble the corner fractures of non-accidental injury. In non-accidental injury the child’s bone texture is normal, and there is no cupping or fraying of the metaphysis where there is a fracture. The appearances of copper deficiency have been described as scorbutic, as in both there is severe osteopenia and spur formation with corner fractures. There is a superficial resemblance but the radiological appearances are quite distinct. In scurvy there is no fraying of the
metaphyses and the epiphyses have a distinctive etched appearance with a sharp outline and 'washed out' centre. Scurvy can also be easily excluded by biochemical tests.

Skull fractures have not been described in infants with copper deficiency, but they are a common finding in non-accidental injury. The presence of normal bone and in particular the presence of normal wrists and knees renders the diagnosis of copper deficiency of sufficient severity to cause fractures unlikely and this can be stated even in the absence of a serum copper concentration. The absence of anaemia and neutropenia and the physical stigmata further make the diagnosis virtually untenable.

Osteogenesis imperfecta

The diagnosis of severe forms of osteogenesis imperfecta is indisputable by a radiologist or clinician. Milder forms of the disease can be difficult to diagnose and often a diagnosis can only be made with time. Most children with mild osteogenesis imperfecta do not present with fractures early in life but tend to fracture between toddling and adolescence.

There is, as yet, no freely available biochemical test for the disease although interesting research is continuing in the fields of collagen chemistry and genetic markers. Until this work produces a test that is freely and quickly available in a general hospital, the diagnosis of osteogenesis imperfecta will have to be made on clinical and radiological grounds.

Considerable genetic heterogeneity is present in osteogenesis imperfecta. Four genetic subgroups have been identified. Within the groups there is a considerable range of severity, and it is this that leads to the potential diagnostic confusion. It is important for both paediatrician and radiologist to actively consider the question of osteogenesis imperfecta in any child suspected of non-accidental injury. The clinical diagnosis of osteogenesis imperfecta rests on the findings of blue sclera in certain cases, a large fontanelle, excessive joint laxity, small stature, and dentinogenesis imperfecta. Some patients with the disease have normal sclera. The clinical manifestations are very variable.

Dentinogenesis imperfecta of osteogenesis imperfecta is best confirmed by a dentist with a particular interest in the subject. Not all pitted teeth are due to it and an expert opinion should be sought in all cases with teeth defects where the diagnosis is in dispute. Many of these cases occur before tooth eruption and dental changes and x-ray radiography is, therefore, of no diagnostic help.

Fractures both accidental and non-accidental can occur with or without bruising over a fracture site. The presence or absence of bruising over a fracture site gives no indication as to the cause of a fracture. The fact that patients with osteogenesis imperfecta may have a tendency to bruise easily is irrelevant in the context of fractures.

A family history of fractures and deafness is often quoted as supportive evidence of the diagnosis of osteogenesis imperfecta and in many cases this is helpful. However, a detailed history of the type of deafness or the way in which the fractures were sustained should be taken. The fact that a relative sustained fractures when falling from a tree or in a motor bike accident does not mean that there is a family history of the disease.

Children with osteogenesis imperfecta may sustain fractures with minimal force but in those with the mild form of the disease these tend to occur when the child is toddling or walking and not confined to a cot. The fractures of osteogenesis imperfecta are mostly in the diaphyses of the long bones. Metaphyseal fractures are rare and when they do occur there is evidence of gross change of the disease. Osteoporosis in mild cases of osteogenesis imperfecta may be mild but some alteration of the trabecular pattern is usually identifiable. Wormian bones are an almost constant finding in osteogenesis imperfecta, and they have a different distribution to those seen as developmental variants. Children who have sustained non-accidental injury are unlikely to sustain further fractures while in care. Where a diagnosis of non-accidental injury is made incorrectly in a child with osteogenesis imperfecta fractures are likely to continue to occur after the child has been placed in care.

It can be seen, therefore, that there are distinguishing features between the three conditions and diagnostic confusion should not arise.

It is vital that non-accidental injury is not over diagnosed. Most paediatricians and radiologists will not make the diagnosis unless they have strong grounds for it and will err on the side of caution if in doubt.

When a case is contested, before court proceedings start, it is essential that the barrister and solicitor are properly briefed, so that they fully understand the medical evidence. Medical evidence is given not to seek retribution for the child's injury but to arrange for proper protection for that child, hopefully with rehabilitation of the family unit. It is difficult for non-medically trained judges and juries to understand the subtleties of radiological changes and the nuances of clinical practice.

Yet it is on this evidence that the outcome of a case often rests and will continue to do so while the
adversarial system of the law continues for such cases.

References

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