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Vitamin D deficiency rickets and allegations of non-accidental injury

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Abstract

Vitamin D deficiency rickets has long been recognized as a cause of fractures and fracture-like appearances in young children. Often seen in the early 20th century, rickets has recently been regarded as uncommon; the radiological appearances, familiar to previous generations, may not be recognized for what they are.

This article reports four children with unexplained fractures initially attributed confidently to non-accidental injury. In each case, the later evidence of vitamin D deficiency led to a reconsideration of that diagnosis.

Conclusion: It is important to be aware of this bone disorder in the differential diagnosis of fractures, to investigate appropriately and to recognize that the radiological appearances may be misleading. A mistaken diagnosis of abuse does real harm, not least to the child itself.

INTRODUCTION

Vitamin D deficiency rickets has long been recognized as a cause of fractures and fracture-like appearances. In the early 20th century when rickets in children was common, the wide variety of clinical features and radiological appearances was well understood. At present, because it seems uncommon in the general population, it may not be recognized immediately for what it is. However, it has recently become clear that vitamin D deficiency is widespread in many populations (1,2).

Vitamin D is obtained in man either from the diet or from synthesis in the skin under the influence of ultraviolet (UVB) radiation, which converts a precursor, 7-dehydrocholesterol, into vitamin D_3. Those most at risk of vitamin D deficiency have limited availability from both sources. These include immigrants in Europe and indigenous communities in a wide range of countries including Turkey, Saudi Arabia and Pakistan. Cases have been reported from North America, particularly in breast-fed infants not receiving supplementation with vitamin D (3,4). In young infants the risk of vitamin D deficiency is related to the nutrition of the mother. If the mother is deficient, the child can have evidence of rickets from the time of birth or before (4,5).

While the finding of multiple fractures in various stages of healing is often regarded as a pointer to non-accidental injury, these can also be caused by heritable and metabolic bone diseases including osteogenesis imperfecta (6), bone disease of prematurity (7) and, more controversially, temporary brittle bone disease (8). This article reports four patients investigated between 1980 and 2000 with good evidence that vitamin D deficiency rickets was the cause of fractures and fracture-like appearances initially attributed to non-accidental injury (Table 1). Seven similar cases have been reported previously (2,9,10).

PATIENTS

Case 1

This girl, born in England of Punjabi immigrant parents, was found to have impaired weight gain after the age of 5 months. She also had microcytic hypochromic anaemia with a low-serum iron, but there was no response to iron therapy. Malabsorption was considered but jejunal biopsy was negative for coeliac disease.

At the age of 34 months, she was again investigated because of continuing poor weight gain. She was found to

Table 1 Clinical features and investigational findings in the four children

Case	Gender	Ethnic background	Age at finding fractures	Predisposing factors for vitamin D deficiency	Principal biochemical findings	Principal radiological findings
1	F	Punjabi	34 months	Probable malabsorption Low intake. Little sunlight exposure	Alkaline phosphatase normal for age	Minor rachitic changes in tibiae, radii and ulnae. Probable pseudofractures of two ribs (Fig. 1)
2	M	Afro-Caribbean	4 months	Breast fed. Probable maternal deficiency	Moderate hypocalcaemia and hypophosphataemia (for age). Grossly raised alkaline phosphatase	Widespread rachitic changes (Fig. 3)
3	M	Caucasian	2 months	Breast fed. Possible maternal deficiency	Raised alkaline phosphatase. Undetectable 25-hydroxyvitamin D	Minor rachitic changes in tibiae, radii and ulnae (Fig. 4)
4	F	Caucasian	3½ months	Breast fed. Probable maternal deficiency	Hypophosphataemia. Grossly raised alkaline phosphatase. Very low 25-hydroxyvitamin D and raised parathyroid hormone	Metaphyseal abnormalities bilaterally in distal femora, proximal tibiae and distal tibiae.

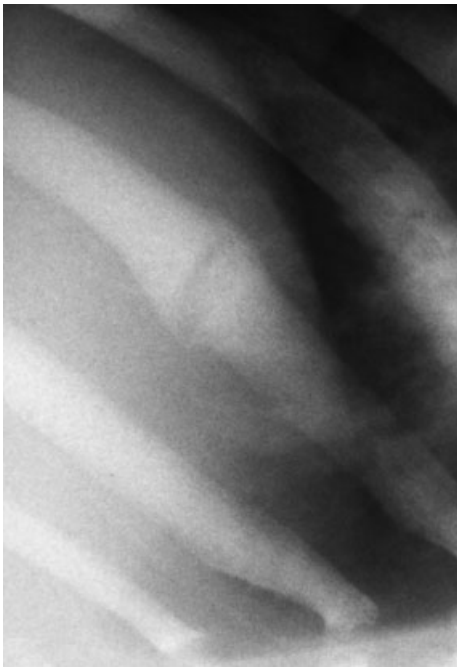

Figure 1 Case 1: fracture-like abnormality of posterior part of the left 10th rib at age 34 months. The appearance of this lesion had not changed in several radiographs over the preceding 4 weeks and was thought to be a pseudofracture.

have a number of bruises and was also reported as having healing fractures of both 10th ribs posteriorly (Fig. 1). Neglect and non-accidental injury were suspected and she was placed in foster care. However, there was no improvement at all in her weight while in foster care. Serial X-rays over 2 months showed no change in the appearance of the rib 'fractures'. At the same time, radiographs of the wrists and knees were reported as showing evidence of a 'minor degree of rickets'. Nutritional review by a dietician indicated a very low intake of vitamin D and little exposure to sunlight. Serum alkaline phosphatase levels were within the reference range for her age. Serum 25-hydroxyvitamin D was not checked at this stage, but she was treated with oral vitamin D. From the records, it is not clear how long this was continued.

She returned to her parents at the age of 41 months, but was re-investigated at the age of 48 months. Bruises were seen, but healing fractures of the left 10th and 11th ribs were also reported; that of the 10th rib was identical in site with the one seen earlier. The radiologist concerned thought that this was, in fact, a pseudofracture (implying rickets) but was asked by the clinician in charge to revise his report. The child was again taken into foster care. After a court hearing at the age of 57 months, she was again returned to her parents.

One factor in this child's rickets was the probable malabsorption for which no cause had been found. Additional factors were that both parents showed evidence of vitamin D deficiency and that an older child had evidence of rickets when admitted to hospital with a fractured femur at the age of 1 year.

Case 2

This boy was born at 36 weeks gestation (2807 g) of Caribbean parents in England. He was breast fed. At the age of 4 months, his parents found a swelling of the left thigh and sought hospital attention. X-rays showed evidence of an oblique fracture of the femoral shaft (Fig. 2). On radiological and clinical grounds, the fracture was recent, consistent with the parents' account, but without superficial evidence of injury.

Elsewhere in the skeleton, there was gross evidence of rickets at the lower ends of both femora, upper and lower ends of both tibiae and fibulae, upper and lower ends of both humeri, the lower ends of both radii and ulnae and the costo-chondral junctions (Fig. 3). The clinical biochemistry showed low levels of serum calcium (1.9 mmol/L) and inorganic phosphate (1.0 mmol/L). The serum alkaline phosphatase was markedly raised at about five times the age-appropriate reference range. Surprisingly, serum 25-hydroxyvitamin D was not measured at this stage. The child's

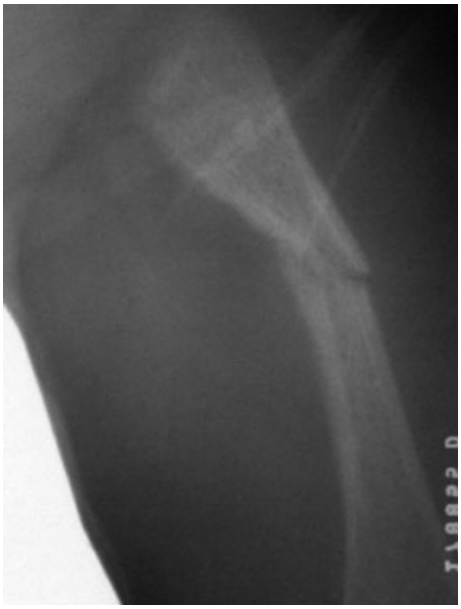

Figure 2 Case 2: left femur to show recent oblique fracture.

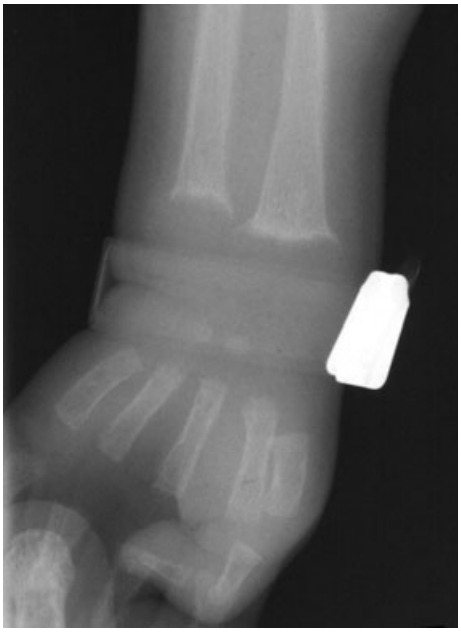

Figure 3 Case 2: right wrist to show rachitic changes in the distal radius and ulna.

mother had symptoms suggestive of osteomalacia but was not investigated at the time.

Following hearings, the child and his siblings were removed from the family, in part, because of other alleged concerns about parenting skills. An appeal was unsuccessful.

Case 3

This Caucasian boy was born at term (4763 g). He was breast fed. At the age of 6 weeks, he had a convulsion. Two weeks later, he had a number of small bruises and was referred to hospital. He was found to have a fracture with callus of the left clavicle. The upper tibiae on both sides and the left upper fibula showed rachitic irregularities (the right upper fibula was not well seen on the films of this date). The lower ends of both tibiae and the distal ends of both radii and ulnae showed minor rachitic changes. The skull showed some widening of the suture lines and the costo-chondral junctions were enlarged. The changes in one upper tibia were reported as representing a metaphyseal fracture (Fig. 4).

Further investigation showed a low-serum calcium (2.0 mmol/L) and raised-serum alkaline phosphatase (about twice the upper limit for his age). 25-hydroxyvitamin D was undetectable in the serum (<3 nmol/L or 1 ng/mL).

A further bruise was found in hospital and attributed to the parents. The radiological view was that the rickets was mild and did not explain the fractures. As a result of a court order, he was discharged into foster care with an aunt. A police investigation was initiated but later dropped. He remained in foster care and the parents appealed for his return. By the time the appeal was heard his mother was seriously ill with leukaemia from which she died. At a court hearing, the care order was revoked, but he remained with the aunt. He had no fractures in the subsequent 20 years.

Case 4

This Caucasian girl was born at term (2977 g). She was breast fed and, at the age of 9 weeks, developed a large red raw area on the neck with adjacent bruising. Treatment for a seborrhoeic dermatitis caused an initial improvement. The parents again sought advice 2 weeks and 4 weeks later because the condition had not improved and bleeding from some lesions had been noted. She was admitted to hospital not only for treatment of what was regarded as seborrhoeic dermatitis with secondary infection but also for evaluation of possible child abuse.

X-rays showed a right parietal lucency interpreted as a fracture. She had healing fractures of the left second, third

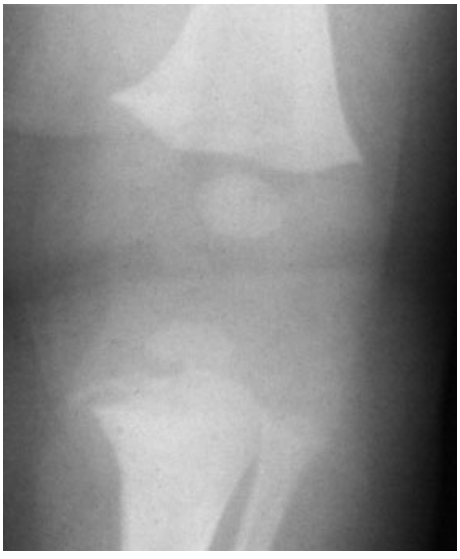

Figure 4 Case 3: left knee to show rachitic changes simulating a metaphyseal fracture in the upper tibia.

and fourth ribs laterally with frayed appearances of the third and fourth anterior rib ends. Multiple abnormalities, all initially regarded as fractures, were reported at both distal femora and both proximal tibiae; a 'fraying of the metaphyses' was regarded as other evidence of trauma. Bilateral metaphyseal abnormalities of the distal tibiae were interpreted as bucket-handle fractures. To the radiologist, these were 'unequivocal findings suggesting repetitive non-accidental trauma'. There was no clinical evidence of any of the fractures and the parents had no recollection of any injury apart from a fall, which might have explained a skull fracture.

Routine biochemical evaluation showed a grossly raised serum alkaline phosphatase and a low-serum inorganic phosphate. The serum 25-hydroxyvitamin D was 9 nmol/L (3 ng/mL). (Levels lower than 50 nmol/L or 20 ng/mL are regarded as indicative of vitamin D deficiency (1).) The serum parathyroid hormone was greatly raised at 298 ng/mL (reference range 12–72 ng/mL).

The parents were told that the child had nutritional rickets, but that the fractures were not consistent with this diagnosis and that non-accidental injury could not be ruled out. Scurvy was suggested by one doctor as the cause of the bruising, but no relevant investigations were undertaken. Evaluation of the mother's diet by a nutritionist suggested a low-vitamin D intake. The child was treated with vitamin D but remained in foster care for 5 months before being returned to her parents. She was well 1 year later.

DISCUSSION

A finding of fractures with rachitic changes in a child's X-rays may be interpreted as evidence of both inflicted trauma (11–13) and neglect. However, rickets is common in certain ethnic groups; it is well recognized even in Caucasian infants particularly if breast-fed (3,4). Fractures have long been recognized consequences of rickets in children (and of osteomalacia in adults). In addition, both in rickets and in osteomalacia, pseudofractures (Looser's zones, Milkman's fractures) are well recognized.

In infants with rickets, it is often difficult to distinguish fractures from pseudofractures. One pointer is that a pseudofracture is not accompanied by the clinical signs associated with a true fracture. Another is that, unless the patient is treated with vitamin D, the radiological appearances of a pseudofracture do not evolve as would be expected of a true fracture. The unchanging appearances of the rib abnormalities in case 1 illustrate this point. However, in rickets, the healing of true fractures may also be delayed (2). If a confident diagnosis of rickets is made, it is perhaps academic to distinguish fractures from pseudofractures, as both are integral features of the condition.

It is not always appreciated that only a minority of infants with significant vitamin D deficiency show radiological abnormalities (14). In addition, the 'paradox of rickets' recognized by early authors is that epiphyseal changes become less obvious with progression of the condition; the classical radiological signs are not seen in a child who is not growing. This issue is illustrated by case 3. While rickets was identified, it was thought on radiological grounds to be mild and insufficient to explain the fractures. At the same time, it was clear from the biochemical findings that the rickets was very severe.

The clinical features of rickets, familiar to previous generations of paediatricians, may now go unrecognized. In case 3, it is likely that the convulsion was the result of the hypocalcaemia and may have caused the clavicular fracture. A convulsion is a common presenting symptom of rickets (3,14). Three of the patients had bruises as part of their presentation. These might be thought to strengthen the likelihood that abuse was the cause of the fractures, but it is also known that vitamin D deficiency rickets can be associated with thrombocytopenia, which is reversible with vitamin D therapy (15). Another possibility is concurrent deficiency of vitamin C (16).

In the diagnosis of vitamin D deficiency rickets, it is important to recognize the limitations of radiology in excluding significant bone disease (2,14). It is also important to be aware that simple clinical biochemistry (serum calcium, inorganic phosphate and alkaline phosphatase) may not be helpful. For example, misleadingly high values of alkaline phosphatase may reflect healing fractures and misleadingly low values may be found in a child who is not growing (as in case 1). Specific assays for serum 25-hydroxyvitamin D and parathyroid hormone are needed.

The small number of case reports of the misdiagnosis of rickets as non-accidental injury may not reflect the true incidence of this problem but rather may reflect the infrequency with which the diagnosis of vitamin D deficiency rickets is considered. As with other disorders contributing to fractures in infancy, a failure to make the correct diagnosis does great harm to the family and, not least, the child itself.

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