Metacarpal morphometry in adults with osteogenesis imperfecta

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Summary and conclusions

Osteogenesis imperfecta is often regarded as a form of osteoporosis. In many cases, particularly those in whom the first fracture occurs outside the neonatal period, bones that have not been fractured may appear radiologically normal. In a group of 24 adults with osteogenesis imperfecta the thickness of the metacarpal cortex was normal but their bones were often slender.

Osteoporosis is probably not an inevitable feature of such cases, and some of the radiological abnormalities reported may be the results of previous fractures and their treatment.

Introduction

Osteogenesis imperfecta is a generic term for a poorly defined group of disorders characterised by abnormal fragility of the bones in childhood. In most patients who have fractures before or at the time of birth the radiological abnormalities are obvious. In others, notably those whose first fracture occurs later, the skeleton may appear normal during the first few years of life. A wide variety of radiological abnormalities are found later in childhood.[1] Some of these abnormalities may not be features of the underlying disease but simply result from previous fractures and the immobilisation required during treatment.

To test this suggestion the metacarpal cortex of adults with osteogenesis imperfecta was measured. The second right metacarpal was chosen because very full information is available on normal adults and because metacarpals are seldom fractured.

Patients and methods

Sixteen women and eight men with osteogenesis imperfecta were studied. Apart from two women and three men all had a family history of the disease and would be regarded as having the benign dominantly inherited disorder. The number of fractures varied from three to over 200. The patients were aged 20 to 70 years.

Standard anteroposterior films of both hands were taken and the dimensions of the second right metacarpal—the outer diameter (D), the diameter of the medullary space (d), and the length (L)—were measured. Derived data included the cortical thickness (D-d), and two of the most widely used indices—$(D^2-d^2)/D^2$ and $(D^2-d^2)/DL$.[2-4] The results were related to normal values for the patient's age and sex[3] and expressed in terms of the standard deviation.

Results

Fig 1 shows that while the cortical thickness (D-d) was normal in the patients with osteogenesis imperfecta, the bones were generally slender; the typical appearance of the metacarpals in osteogenesis imperfecta is shown in fig 2. In most of the patients the length of the metacarpals was normal. Fig 3 shows that $(D^2-d^2)/D^2$ was abnormal, the high value reflecting the slenderness of the bone rather than any abnormality in the cortical thickness. The metacarpal index, $(D^2-d^2)/DL$, was normal in most patients.

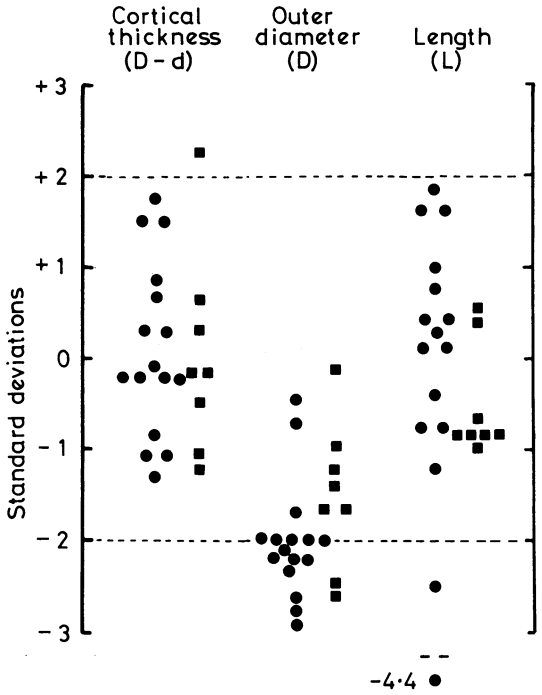

FIG 1—Cortical thickness, outer diameter, and length of second right metacarpal in adults with osteogenesis imperfecta. D = Outer diameter. d = Diameter of medullary cavity. L = Length. ● = Women. ■ = Men. Each point represents standard deviation from mean for a normal population of same age and sex.[3]

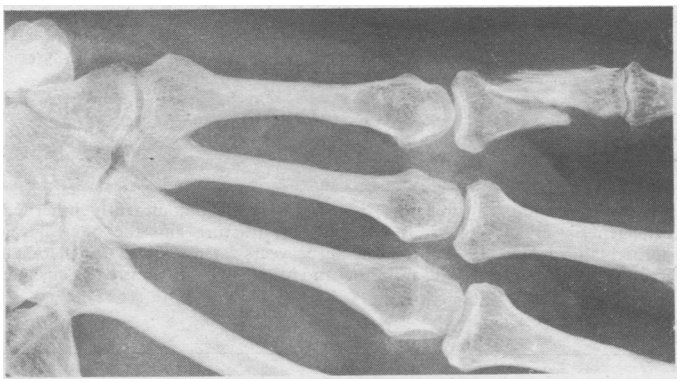

FIG 2—Part of hand radiograph from 42-year-old woman with dominantly inherited osteogenesis imperfecta. Metacarpals are slender but cortical bone has normal thickness.

Discussion

Thin cortical bone is often regarded as a radiological feature of osteogenesis imperfecta,[5-8] but these results show clearly that a thin metacarpal cortex is not found in affected adults. Radiographs of other parts of the skeleton were available in many cases

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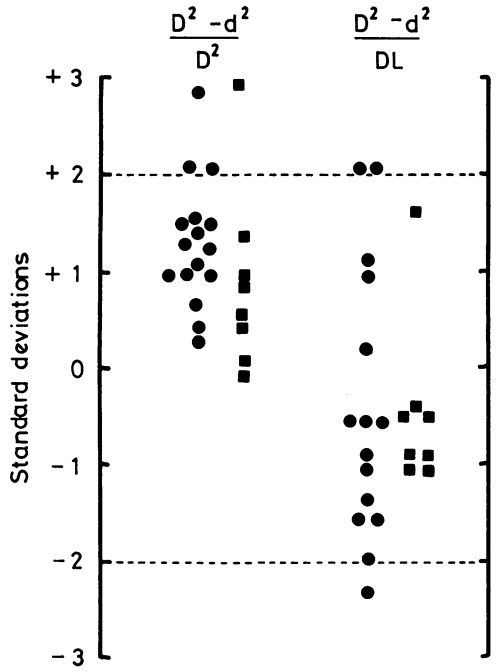

FIG 3—Derived indices of bone mass in adults with osteogenesis imperfecta. Symbols and expression of results are the same as those for fig 1.

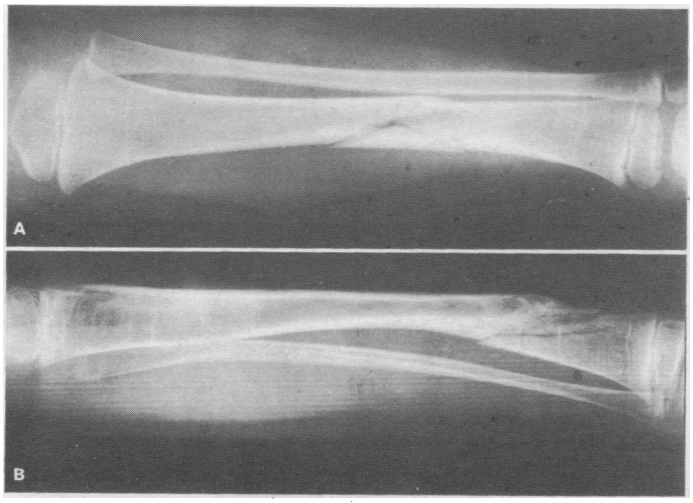

FIG 4—(*a*) Fractured tibia in 3-year-old boy with dominantly inherited osteogenesis imperfecta. This was his first fracture and the bone has a normal appearance. (*b*) Same bone three years later at time of its fourth fracture. Bone appears quite abnormal and muscle mass is greatly diminished.

and much-fractured leg bones almost always had thin cortices. Many of the textbook descriptions of the radiology of osteogenesis imperfecta tarda or dominantly inherited osteogenesis imperfecta probably reflect simply the ravages of previous fractures. A common feature of the history in many cases is that one limb has been the site of many fractures. Such a limb has become particularly vulnerable, presumably because osteoporosis, due to repeated immobilisation, has been superimposed on the underlying bone disease.

Individual reports of patients with apparently normal bones have been made before.[9 10] The fragility of radiologically normal bones is at first sight surprising, but recent work has made it increasingly clear that in osteogenesis imperfecta the abnormality is not in the mineral component of bone but in the organic matrix.[11] Defects in the composition or cross-linking of bone collagen might seriously affect bone strength without affecting radiological appearances.

The normal appearances of bone in the early years of life are illustrated in fig 4. Bone may appear normal when the first few fractures occur, since the problems of differential diagnosis are most difficult at this stage. In particular, a child with unexplained fractures may be thought to have suffered non-accidental injury,[10 12] and a mistaken diagnosis of abuse may have serious consequences.

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SHORT REPORTS

Epilepsy in Paget's disease of the skull

Paget's disease of the skull has several different neurological complications including headache, deafness, optic atrophy, basilar invagination with compression of the hind brain, and hydrocephalus with dementia.[1 2] We have been unable to trace a reference to epilepsy as a complication, and we think the following two histories illustrating the association of the two disorders are interesting.

Case reports

Case 1—An Englishman aged 52 presented in March 1976 giving a three-month history of three attacks of loss of consciousness. His wife had witnessed the attacks and described mouth drooping, head shaking, eyes rolling, and generalised involuntary movements. On regaining consciousness he complained of severe headache and tiredness but was not aware he had had the attack. He had suffered from migraine until two years before, but there was no previous or family history of epilepsy. Results of investigations were as follows: nervous and cardiovascular systems normal; blood pressure 140/80 mm Hg; skull radiography, a localised area of osteoporosis in the left frontotemporal region (see figure); electroencephalography, focal sharp