Discussion

Professor J. Spranger: The Fairbank type of cystic osteogenesis imperfecta seems to differ from the cases you described. Do you have earlier films of these four patients?

Professor Dent: No, I don't. Roger Smith (Smith, Francis and Bauze, 1975) reported a rather similar case, and claimed that the earlier films did not show these curious rounded structures that I have demonstrated in the humeri. I have not contacted him to verify the presence or otherwise of the other very different features described in Fairbank's cystic type of osteogenesis imperfecta which has cysts all over the place. The humeral lesions seem to be a gross Paget-like change, and I think the condition could be juvenile Paget's disease. Smith had seen only one case and so have I, so that it must be extremely rare.

Dr P. Jacobs: I have heard it said that liver disease is the commonest cause of osteoporosis in children. Is this true? Also why does osteoporosis occur with biliary atresia?

Professor Dent: Liver disease cannot possibly be the commonest cause in children. For one thing, liver disease of that severity is not very common, and it must be very severe to cause osteoporosis. Regarding your second question, I will refer you to Dr Stamp who, when with me, worked on the problems of liver disease and its effect upon vitamin D and calcium metabolism.

Dr T. C. B. Stamp: Severe liver disease in a child may lead either to osteoporosis or rickets and osteomalacia. The simplistic explanation is that although with osteoporosis there is a very high calcium requirement which may exceed the child's calcium intake, there is no associated deficiency in vitamin D. In osteomalacia, vitamin D may be deficient owing to malabsorption, or to vitamin D loss if there is a biliary fistula interfering with the circulation of the vitamin and its metabolites. Finally, there is the more theoretical possibility of a defect of 25-hydroxylation in severe obstructive liver disease.

Professor Dent: There may be gross osteoporosis in children with complete biliary atresia, but no rickets in the cases that I have seen.

Dr I. R. S. Gordon: What radiological signs distinguish between osteomalacia and osteoporosis if the child is not actively growing?

Professor Dent: Well, of course there may be rickets which sometimes has secondary hyperparathyroidism which may show in the fingers; but if the child has stopped growing there will be awful difficulty. Should the condition continue long enough there will be smooth bicconcave vertebral bodies, whereas in osteoporosis they tend to be irregularly fractured and collapsed. If fractures should appear after the cessation of growth, new bone may form quickly although it doesn't calcify but forms a Looser zone. But in occult osteomalacia, presenting with myopathy and before there are any bi-concave vertebral bodies, Looser zones or hyperparathyroidism, there may be no distinguishing radiological signs.

Professor Spranger: May I return to osteogenesis imperfecta and the cystic type described by Fairbank. I think that there are sometimes survivors of the thick bone recessive type; but when one of these patients survives, they eventually develop the cystic form. Amongst the thin bone type there are both dominant and recessive varieties, so there may be three or even four types. Do you distinguish between the congenita and tarda forms? For example, the thin bone type may be congenital.

Professor Dent: The term 'congenita' to me means only present at birth. It does not indicate whether the condition is genetic or not, nor dominant or recessive. There are certainly several forms of the congenita type, so that this term needs qualification, e.g. thick bone or slender bone type.

Dr P. S. Harper: I am very unconvinced that there are all these different types of osteogenesis imperfecta; and apart from the South African cases I am unsure about the existence of recessive transmission except for a very small number. Do you think that there is any good evidence to support this separation?

Professor Spranger: In the thick bone type, I have never seen vertical transmission. The parental age is normal and there are numerous instances of affected siblings. In Algeria there is a whole population group with heavy consanguinity, and there were two cases reported in the American Journal of Diseases of Children who were the offsprings of consanguinous parents. They were all of the thick bone type. There has not been a single indication that the thick bone type could be due to a dominant transmission. It proves also that if one child in a family has the thick bone type, so do the others. Not a single instance is known of thick and slender bone types being manifest in the same sibship. Dominant transmission of osteogenesis imperfecta is well known, but there are several recent series, including reports from Switzerland where the patients, offsprings of consanguinous parents, had only the slender bone type, everything being consistent with recessive transmission of this type too. The thick bone type is certainly recessive: I also think that there are two forms of the slender bone variant.

Professor Dent: Yes, I entirely agree. Genetic disease usually displays a good deal of phenotypical variation, as in sickle-cell anaemia. Moreover, this free discussion promotes useful practical knowledge to help us advise our patients about the prognosis. I think that the view we have suggested is a sound working hypothesis for osteogenesis imperfecta.

Dr Harper: I agree in general, but remain to be convinced in particular.

Professor Spranger: Well, once it is shown in a single case that the thick bone type does not breed true, you will be right of course.

Reference

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