Coeliac disease can still present with osteomalacia!

Sir, Coeliac disease is known to be associated with an increased prevalence of osteoporosis, although the exact mechanism of this is uncertain. However, osteomalacia is now rarely reported in association with coeliac disease in the Western world. We describe three cases of osteomalacia presenting in middle-aged Caucasian women, all of whom had previously undiagnosed coeliac disease.

A 60-yr-old lady with insulin-dependent diabetes presented with a 15-yr history of diarrhoea and low back pain. She was noted to be only 140 cm tall and weighed just 40 kg. Initial investigations revealed a macrocytic anaemia with a raised alkaline phosphatase and a corrected calcium of 2.01. Her diarrhoea had been previously investigated by a number of specialists and a structural cause excluded. She had been found to be folate deficient but had failed to respond to oral supplements. Direct enquiry revealed that she had not walked unaided until the age of 6 yr. Examination showed a marked kyphoscoliosis, prominent costal margins and spinal tenderness with proximal muscle weakness. Radiological examination demonstrated Looser’s zones with thoracic vertebral collapse. Autoantibodies were all negative, but a small bowel biopsy demonstrated total villous atrophy. Treatment with a gluten-free diet, bisphosphonates and supplemental calcium and vitamin D led to a gradual resolution in her symptoms and a marked improvement in small bowel histology at 6 months.

A 54-yr-old woman presented with 1 yr of increasing proximal muscle pain and weakness, together with spontaneous pain in the thoracic spine. She denied any other symptoms or past medical history and went through the menopause at the age of 47 yr. Examination showed her to be 155 cm in height and weighing 52 kg. She had a kyphosis with thoracic spinal tenderness and proximal muscle weakness. Radiographs showed fractures at T12, L4 and L5, together with Looser’s zones in the pubic rami. Bone chemistry showed an elevated alkaline phosphatase of 234 and a reduced calcium of 2.02. Full blood count, electrophoresis and thyroid stimulating hormone were normal. Her anti-endomysial antibody test was positive. A diagnosis of osteomalacia was made and an endoscopic duodenal biopsy undertaken. This demonstrated near total villous atrophy. Treatment with bisphosphonates, calcium and vitamin D were given, together with a gluten-free diet. A repeat duodenal biopsy showed a dramatic improvement 6 months later and her symptoms gradually resolved.

A 43-yr-old pre-menopausal woman complained of pain in her spine and proximal musculature for 1 yr. She was otherwise asymptomatic and was of normal build. Examination initially suggested fibromyalgia but bone chemistry revealed a raised alkaline phosphatase and a slight low corrected calcium (2.13). An anti-endomysial antibody test was positive and plain films showed osteoporotic vertebral collapse. An isotope bone scan showed widespread increased uptake consistent with osteomalacia. Endoscopic small bowel histology confirmed subtotal villous atrophy and treatment with a gluten-free diet and supplemental vitamin D was commenced. Symptomatic improvement occurred promptly and small bowel histology had normalized at 4 months.

An association between coeliac disease and osteomalacia was first reported in 1953 [1]. Recognition that this could occur without clinical features of malabsorption followed [2]. It was reported that osteomalacia could be the presenting feature of coeliac disease [3, 4] and that vitamin D supplementation was an effective mode of therapy [5]. However, a recent paper highlighted the fact that coeliac disease now frequently presents with tiredness and anaemia, rather than the consequences of malabsorption [6]. Most clinicians now rarely see clinical or biochemical evidence of osteomalacia in patients with coeliac disease and two recent large series of coeliac patients reported no evidence of osteomalacia [7, 8]. Nonetheless, three patients with osteomalacia were identified among 21 newly diagnosed coeliac patients in a study by McFarlane et al. [9] and the improvement in bone density with treatment was greater in these three patients than in the remainder who had varying degrees of osteopenia. A combination of a gluten-free diet and vitamin D supplementation proved very effective.

We have described three cases of osteomalacia presenting as a major feature of coeliac disease in middle age. These patients all had clinical and radiological features of osteomalacia which pre-dated their diagnosis by a significant time period, and two also had other features of malabsorption. Untreated coeliac disease is associated with malabsorption. Fat-soluble vitamins are generally absorbed in the distal small bowel, but extensive villous atrophy from long-standing gluten enteropathy may lead to poor absorption of vitamin D [10] and other vitamins.

Serological tests for coeliac disease have increasingly become both more sensitive and specific. The anti-gliadin antibody test carries a high sensitivity and speci-
ficity, but has been superseded in most laboratories by testing for the anti-endomysial antibody which is over 90% sensitive and nearly 100% specific in younger patients [11]. This has been advocated as a screening test for coeliac disease in all young patients with osteoporosis [12]. However, there is evidence that this may be less useful in screening older populations who may present with a more selective isolated iron deficiency picture [13]. Radiological evidence of osteomalacia has become increasingly rare in Western Europe and is a late feature of vitamin D deficiency. However, radiological evidence of osteoporosis in the form of vertebral collapse was invariable in our patients, and low impact fractures of the hip or pubic rami should alert the clinician to the possibility of associated early osteomalacia.

The diagnosis of coeliac disease should be suspected in any patient with osteomalacia, although the classical biochemical features of osteomalacia occur late in the process of vitamin D depletion and earlier features include an increase in parathyroid hormone as well as a reduction in 25-hydroxy cholecalciferol levels. These clues may also be worth seeking in patients with severe unexplained osteoporosis, particularly where anaemia is an associated feature. Clearly a gluten-free diet is a mandatory part of therapy in coeliac disease. The treatment of associated bone disease might also include the use of parenteral vitamin D supplements with or without bisphosphonates.

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