CASE REPORT

Suspected coexistent osteoporosis and osteomalacia with atraumatic bilateral neck of femur fractures in a 53-year-old man without apparent risk factors

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SUMMARY

Atraumatic bilateral hip fractures in the relatively young are exceedingly rare. In this case report, we present one such patient diagnosed by MRI and treated with bilateral hip screws. Subsequent investigations revealed severe osteoporosis and primary 25-hydroxyvitamin D (25OHD) deficiency at a level suggestive of concurrent osteomalacia.

BACKGROUND

This case first highlights the importance of employing MRI where doubt exists with vague hip pain. Second, it emphasises that osteoporosis may occur in those without typical risk factors. Finally, it highlights the contribution of 25-hydroxyvitamin D (25OHD) deficiency to bone demineralisation, as well as its role in the pathogenesis of osteomalacia, which may be an underappreciated disease.

CASE PRESENTATION

A 53-year-old Irish man presented to the emergency department with left-sided groin pain, which had started insidiously 4 weeks prior. An X-ray taken at this presentation showed no abnormalities and he was discharged home. He represented a further 4 weeks later with ongoing pain and was referred to the orthopaedic service. The patient's medical history included mild hypertension and hypercholesterolaemia. He denied any history of chronic back or joint pain, previous fractures or recent trauma. On examination the patient presented with left-sided groin pain with active straight leg raising and passive rotation of the left hip. Right hip examination was normal. There were no gross physical abnormalities such as kyphosis, stooped posture, leg length discrepancy or joint deformities. Repeat X-ray was normal, showing no fractures, space occupying lesions or bony lesions.

INVESTIGATIONS

MRI showed bilateral undisplaced, intracapsular femoral neck fractures (figures 1 and 2). The patient underwent bilateral dynamic hip screw stabilisation of the fractures. Postoperatively, the patient reported significant improvement in groin pain. Bone reamings obtained intraoperatively showed no evidence of malignancy. Further investigation revealed osteoporosis with a T-score of -2.8 at the lumbar spine. Serum 25OHD analysis showed severe deficiency 8.3 nmol/L (3.3 ng/mL). Given the severely deficient serum 25OHD level,

concurrent osteomalacia was suspected. However, the patient did not report pain or weakness prior to the four-week history on first presentation, nor were there any suggestive findings on clinical exam such as throbbing bone pain on pressing on his sternum or long bones. He had no history of myositis, fibromyalgia, chronic fatigue syndrome or any other conditions that can mimic osteomalacia. Biochemical bone profile, endocrine function and renal function were all measured and were found to be within normal limits (table 1). He denied any history of chronic back or joint pain, previous fractures or recent trauma.

Despite having a normal biochemical liver profile, alcohol intake was reported at 400 g/week (recommended maximum 170 g/week¹). He was overweight (body mass index 28.9 kg/m²) and led a generally sedentary lifestyle without any major physical activity socially or at work. Dietary evaluation did not immediately reveal an imbalanced or restrictive dietary pattern, nor did he report unusually low sunlight exposure.

TREATMENT

The patient was referred to both the Clinical Nutrition and Endocrinology services, and oral vitamin D replacement therapy was started

Figure 1 T1-weighted MRI of both hips showing an irregular line of decreased signal through both femoral necks in keeping with fracture.

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Figure 2 T2-weighted MRI of the same hips showing increased signal around the femoral necks in keeping with acute fractures.

according to the American Endocrinology Guidelines (6000 IU/day).²

OUTCOME AND FOLLOW-UP

At 6-month follow-up he was pain free, X-ray showed no evidence of previous fracture, and serum 25OHD had increased to 124 nmol/L (49.6 ng/mL).

DISCUSSION

This case was exceptional for three broad reasons. First, presentation by a relatively young man with atraumatic bilateral hip fracture is extremely unusual. While hip fracture risk increases

exponentially with age secondary to decreased bone mineral density,³ hip fractures in the young usually follow high-energy trauma. Fracture following minimal trauma is usually identified in patients with metabolically altered bone states^{4, 5} including hyperparathyroidism, chronic steroid use and chronic malabsorptive conditions, none of which applied in this case.

Second, the patient was subsequently diagnosed with osteoporosis of the lumbar spine based on dual X-ray absorptiometry T-score of -2.8 , although biochemical indices were not abnormal (table 1). The assessment was not conducted at the hip due to the fractures and surgical intervention, but osteoporosis in that region must be assumed given the clinical scenario. Excepting the high-alcohol intake and 25OHD deficiency, this 53-year-old man did not have significant additional risk factors for osteoporosis. There was no endocrine imbalance, malabsorption, malnutrition or steroid use. This therefore highlights the contribution of 25OHD deficiency to osteoporosis, as well as the insidious nature of bone demineralisation in an apparently healthy man. This case emphasises that osteoporosis should not be dismissed by clinicians as part of normal age-related decline, but should be regarded as an independent disease entity. Orthopaedic surgeons are in a unique position to identify and treat osteoporosis.⁶

Finally, this case was noteworthy due to the exceptionally low 25OHD of a level at which osteomalacia might feasibly occur. Serum 25OHD deficiency is more prevalent than previously thought.⁷⁻⁹ Up to 40% of the Irish adult population have insufficient serum 25OHD, with 6.7% having severe deficiency of less than 30 nmol/L (12 ng/mL).⁷ Aetiology is varied, but includes primary deficiency from lack of natural sunlight and poor diet. However, severe deficiency is almost always caused by malabsorption.¹⁰ Serum 25OHD deficiency and associated secondary hyperparathyroidism result in increased phosphate loss. The resulting imbalance between free calcium and free phosphate causes defective mineralisation of the bone matrix.¹¹ Serum calcium and phosphate levels are often normal as a result of this feedback mechanism, as was the case in this patient (table 1). In children, this mechanism is known as rickets, and is characterised by the bony, rachitic, bowed legs caused by the force of the child's weight on poorly mineralised matrix. In adults, the equivalent condition is osteomalacia, and is not accompanied by the same long bone deformities. Instead, it may present with throbbing bone pain that may be elicited by pressing a forefinger or thumb against the sternum, tibia or midshaft of the radius or ulna.¹¹ While the pathophysiology of bone pain is unknown, it likely results from poor structural support of the periosteal covering and its sensory nerve fibres. It can often be mistaken for myositis, fibromyalgia or chronic fatigue syndrome. Treatment with oral vitamin D according to the guidelines resulted in resolution of 25OHD deficiency. At 124 nmol/L (49.6 ng/mL) the values were well above the recommended levels, but were not in the toxic range. The USA Endocrine Society suggested that an upper limit of 250 nmol/L (100 ng/mL) for 25OHD be used.²

Clinical, biochemical and radiological findings do not reliably differentiate osteoporosis and osteomalacia, and in fact, both can coexist. Bone biopsy with prior double-labelling (rarely conducted outside of specialised clinical or research centres) is required for absolute diagnosis, however, in light of the insufficiency fractures in this case, along with extreme vitamin D deficiency and normal bone profile, osteomalacia is the most probable underlying aetiology. In this case, the clinical situation was further complicated with a diagnosis of osteoporosis of the lumbar spine based on dual X-ray absorptiometry T-score of

Table 1 Comparison of biochemical, endocrine and renal values to institutional laboratory reference ranges

Investigation	Value	Institutional reference range
Biochemical bone profile		
25OHD, nmol/L (ng/mL)	8.3 (3.3)	>50 (20)
Calcium, mmol/L	2.27	2.2–2.6
Phosphate, mmol/L	1.11	0.8–1.5
Magnesium, mmol/L	0.8	0.7–1.0
Albumin, g/L	33	35–50
Alkaline phosphatase, IU/L	134*	30–130
Endocrine function tests		
Parathyroid hormone, pg/mL	54	15–65
Free T4, pmol/L	14.5	7.0–16
Follicular stimulating hormone, mIU/mL	4.6	1–12
Luteinizing hormone, mIU/mL	0.9	1–9
Androstenedione, nmol/L	0.6	0.5–10.1
Renal profile		
Urea, mmol/L	3.6	2.5–7.8
Sodium, mmol/L	138	133–146
Potassium, mmol/L	3.9	3.5–5.3
Chloride, mmol/L	100	95–108
Creatinine μ mol/L	66	64–104

All biochemical tests were conducted by the Medical testing Laboratory of Beaumont Hospital, Dublin, Ireland, an accredited laboratory (Irish National Accreditation Board). 25OHD, 25-hydroxyvitamin D.

–2.8. It is worth noting some inherent limitations in dual X-ray absorptiometry. T-scores are derived from bone mineral density which in turn is a function of mineral and collagen composition. In the presence of osteomalacia, total collagen can be increased but the decrease in mineralisation of this new collagen leads to a decreased bone mineral density which makes it difficult to decipher a diagnosis of osteoporosis in the presence of osteomalacia without a bone biopsy. Bone densitometry has been shown to detect osteoporosis in only 70% of patients with osteomalacia.¹² However, despite these limitations, the profoundly negative T-score in the presence of insufficiency fractures with no other risk factors is strongly suggestive of coexistent osteoporosis.

In the present case, as with the osteoporotic state, there were no obvious causes of vitamin D deficiency. Risk factors include malabsorption, poor diet and lack of sunlight. Vitamin D as a fat-soluble vitamin is stored in body fat and obesity reduces the bioavailability of cholecalciferol by sequestering it in deep body fat.¹¹ Vitamin D is also affected by corticosteroid levels, which can be mildly elevated with increased alcohol content, affecting both osteoporosis and available vitamin D.¹³ However, while overweight, this patient was not obese, not diabetic or prediabetic and was found to have normal circulating corticosteroid levels. Evaluation by the Clinical Nutrition, Endocrine and Gastroenterology services found no restrictive or unbalanced dietary pattern, nor was there any evidence of malabsorption. The patient did not report unusually low sunlight exposure. Excess alcohol intake and alcohol dependence are associated with malnutrition and poor diet, but no direct effect on vitamin D levels has been definitively proven. While these factors may have been contributory, they do not explain the degree of deficiency.

In conclusion, we present a 53-year-old man with atraumatic bilateral hip fractures that were diagnosed by MRI and not

evident by X-ray. Although he did not have typical risk factors, he was diagnosed with osteoporosis of the lumbar spine. Severe 25OHD deficiency was also evident which was likely to contribute to the osteoporotic state and ultimate fractures and may have resulted in concomitant osteomalacia, although this could not be definitively ascertained in the absence of bone biopsy. This case report highlights the value of obtaining an MRI to complement X-ray evaluation where pain persists, and in particular highlights the importance of measuring serum 25OHD levels so that the appropriate medical therapy, supplementation and dietary intervention may be established without delay.

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Competing interests None.

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Learning points

- In cases where doubt exists with vague or representing hip pain, MRI is required for definitive diagnosis in the case of occult fractures of the neck of the femur.
- Osteoporosis may occur in those without typical risk factors.
- 25-hydroxyvitamin D deficiency contributes to bone demineralisation, as well as the pathogenesis of osteomalacia, which is an underappreciated disease.

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