

Life thru a lens

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CASE REPORT

A 36-year-old man was referred with a four-month history of bone pain that was located in his left hip, lower back and ribs. At the same time he had difficulty in walking partly due to the pain and partly due to muscle weakness of his proximal extremities. His medical history mentioned congenital cataract due to aniridia, for which he received intraocular lens insertions. He was not on any medication. Physical examination showed a pale man wearing sunglasses. He had a prominent kyphoscoliosis. He reported pressure pain between the first lumbar and first sacral vertebrae and all movements of his extremities were painful. Further physical examination was normal. Neurological examination was also normal except for a slight paralysis of both ileopsoas muscles. Magnetic resonance imaging of the spine showed multiple degenerative discs. X-ray of the hip joint was normal. Bone scintigraphy showed multiple osseous abnormalities, mainly in the ribs, left pubic bone and both sacroiliac (SI) joints (*figure 1*), which were suspicious for bone metastases of an unknown primary malignancy. Therefore, computer tomography (CT) scanning of the thorax and abdomen as well as laboratory tests were performed.

Figure 1. Multiple osseous abnormalities, mainly in the ribs (left panel) and in the left pubic bone and both SI joints (right panel)



CT imaging was completely normal, without obvious abnormalities in the vertebrae or other osseous structures. Laboratory tests showed a normal erythrocyte sedimentation rate (6 mm), slight leucocytosis ($11 \times 10^9/l$), decreased calcium (2.08 mmol/l), normal albumin (37 g/l) decreased phosphate (0.58 mmol/l), and increased alkaline phosphatase (415 U/l).

WHAT IS YOUR DIAGNOSIS ?

See page 144 for the answer to the photo quiz.

DIAGNOSIS

Altogether the laboratory tests and bone scan suggested a possible diagnosis of osteomalacia secondary to vitamin D deficiency, which was confirmed by an increased parathyroid hormone (84 pmol/l), and decreased 25(OH) vitamin D (12 nmol/l).

When asked specifically, the patient revealed that he suffered from severe photophobia due to the aniridia and had been avoiding daylight exposure for the past 20 years: he worked nightshifts, slept during the day and always kept his curtains closed. His food habits mentioned no dairy products or fish.

Osteomalacia is a disorder of decreased mineralisation of newly formed bone matrix.¹ Several different disorders cause osteomalacia, but vitamin D deficiency is the most common cause. Severe and prolonged vitamin D deficiency results in hypocalcaemia, secondary hyperparathyroidism, and hypophosphatemia, ultimately causing osteomalacia.² Moreover, vitamin D deficiency is associated with cardiovascular risk factors such as arterial hypertension, diabetes mellitus, chronic kidney disease and dyslipidaemia.³

Clinical manifestations of osteomalacia include diffuse bone pain, polyarthralgias, proximal muscle weakness and difficulty in walking.^{1,4} The diagnosis is based on a combination of clinical manifestations, biochemical tests, radiological features such as pseudofractures (Looser zones) and, rarely, bone histomorphometry.^{1,4}

Vitamin D deficiency should be corrected by supplementation of ergocalciferol (vitamin D₂) or cholecalciferol (vitamin D₃) 50,000 IU (1.25 mg) once a week for eight weeks, followed by dose adjustments based on serum 25-hydroxyvitamin D and PTH levels.¹

The extreme photophobia causing the vitamin D deficiency was due to congenital aniridia. Aniridia is a rare panocular disorder (incidence 1:64,000-1:100,000)^{5,6} causing cataract, glaucoma and nystagmus and is due to mutations in the PAX6 gene on band p13 of chromosome 11.⁵ Iris hypoplasia is the most obvious sign and leads to photophobia.

REFERENCES

1. Bhan A, Rao AD, Rao DS. Osteomalacia as a result of vitamin D deficiency. *Endocrinol Metab Clin North Am.* 2010;39(2):321-31.
2. Ashwell M, Stone EM, Stolte H, et al. UK Food Standards Agency Workshop Report: an investigation of the relative contributions of diet and sunlight to vitamin D status. *Br J Nutr.* 2010;104(4):603-11.
3. Pilz S, Tomaschitz A, März W, et al. Vitamin D, cardiovascular disease and mortality. *Clin Endocrinol (Oxf).* 2011;75(5):575-84.
4. Gifre L, Peris P, Monegal A, et al. Osteomalacia revisited: a report on 28 cases. *Clin Rheumatol.* 2011;30(5):639-45.
5. Lee H, Khan R, O'Keefe M. Aniridia: current pathology and management. *Acta Ophthalmol.* 2008;86(7):708-15.
6. Adeoti CO, Afolabi AA, Ashaye AO, Adeoye AO. Bilateral sporadic aniridia: review of management. *Clin Ophthalmol.* 2010;4:11085-9.