Clinical picture

Profund vitamin D deficiency in coeliac disease and pregnancy

Case presentation
A 23-year-old Turkish woman with known coeliac disease presented to hospital with a fractured left femur (Figure 1 a) sustained when arising from bed. Two years prior, she sustained a fracture at the right humerus (Figure 1 b), caused by lifting the younger of her two children. Hypercalcemia was documented at the time of the first fracture, but she defaulted from follow-up.

Investigation on admission for the femoral fracture revealed iron-deficiency anaemia and undetectable vitamin D levels (in keeping with suspected non-compliance with a gluten-free diet). Her serum calcium was 2.93 mmol/l (normal range [NR] 2.2–2.6) with a serum parathyroid hormone level (PTH) of 29.6 pmol/l (NR 1.1–6.9) and a urine calcium excretion of 10.9 mmol in 24 hrs (NR 2.5–7.5), diagnostic of primary hyperparathyroidism. In addition to her displaced left femoral fracture, extensive lytic deposits were seen throughout the skeleton with one in the left tibia, thought to be at imminent risk of fracture. Ultrasound of the neck was suggestive of a left lower parathyroid adenoma.

She was treated initially with intravenous saline and high-dose parenteral vitamin D replacement, followed by intramedullary nailing of both the left femoral fracture and of the left tibia (prophylactically, on account of incipient fracture). Extensive bone biopsies were taken to exclude malignant bone filtration. Histological examination of all samples gave results consistent with osteomalacia and hyperparathyroid bone disease, with no evidence of malignancy.

One month later, she underwent an uneventful left lower parathyroidectomy; histology confirmed a parathyroid adenoma. Her serum calcium has remained normal over a 2-year period.

Discussion
This is a dramatic presentation of severe metabolic bone disease, with long bone fracture in a young patient after minimal or no trauma, in the context of a ‘perfect storm’ of untreated coeliac disease, a parathyroid adenoma, and two recent pregnancies.

In healthy women, intestinal calcium absorption doubles in early pregnancy to meet the demands of the developing fetal skeleton. Malabsorption of vitamin D would have significantly impaired this in our patient. Lactation is associated with an obligatory calcium loss irrespective of maternal calcium intake. In healthy women, this ‘calcium debt’ is repaid after cessation of breast feeding, but in our patient, poor calcium absorption would have severely limited this. The images presented here
serve as a striking illustration of how underlying coeliac and parathyroid disease, when combined with the changes in calcium metabolism associated with normal pregnancy, can have catastrophic consequences.

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Reference