Multiple Pathological Fractures In A Young Woman
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A 20yr old Turkish female presented to an outside hospital in 2006 with a pathological fracture of her right humerus. Subsequent investigations, arranged because of chronic ill-health and difficulty maintaining her weight, led to a robust diagnosis of coeliac disease (positive serology and classical duodenal histology) but dietary compliance was suboptimal and she was lost to follow-up. She had 2 pregnancies in 2004 and 2007.

Having been lost to follow-up she represented 2 years later (2008), with a left leg DVT, widespread bone pain, poor mobility, weight loss and a left-sided neck mass. Serum biochemistry showed a corrected calcium 2.68mmol/l, phosphate 0.67mmol/l, ALP 1900 iu/l, PTH 413pg/ml (reference range: 5-50); vitamin D levels were not measured. Ultrasound of the neck demonstrated a soft tissue mass (3.6 x 1.5cm) at the lower pole of the left lobe of the thyroid, suggestive of a parathyroid lesion. She was referred for parathyroidectomy, but again was lost to follow-up.

Aged 23, she presented to a second outside hospital with a pathological fracture of her left femur. Serum biochemistry confirmed increasing hypercalcaemia (2.93mmol/L), and undetectable levels of 25-OH Vitamin D. Xrays of the pelvis and lower limbs confirmed widespread lytic lesions. She underwent therapeutic intramedullary nailing of the left femur and prophylactic nailing of the left tibia. Bone histology from the femur and tibia reamings was reported as normal.

The patient was transferred to our institution for further assessment. Bone density imaging confirmed osteoporosis (T-score -3.3 at the lumbar spine, -4.7 at the femoral neck). FDG PET scan showed multiple mixed lytic and sclerotic lesions throughout the skeleton. MRI of the pelvis revealed low signal throughout the iliac bones and proximal femora, with maintenance of the bone cortices. She developed pain in the right femur indicative of imminent pathological fracture and underwent prophylactic intramedullary nailing of the right femur, during which further samples of bone were obtained. Histology on undecalcified bone samples confirmed osteomalacia and no evidence of malignant disease. Following intensive treatment with high dose vitamin D (ergocalciferol), in conjunction with commencement of a gluten-free diet, the patient underwent removal of the left parathyroid adenoma (2 x 1cm) in September 2009. Her most recent serum calcium is 2.15 mmol/L.

This case emphasises the dramatic metabolic bone disease associated with coeliac disease due to vitamin D deficiency complicated by an associated parathyroid adenoma and questions the underlying cause of hyperparathyroidism- whether primary or tertiary. This report also highlights the risk of vitamin D deficient bone-induced complications developing in pregnancy.