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Acid-Base Balance-Electrolyte Quiz – Case 12

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A 61-year-old woman reported progressively worsening bone pain in the left right, lower back and arms over a period of several months. A bone survey revealed generalized osteopenia. Laboratory investigation showed only hypophosphatemia (serum Po_4^{3-} 1.1 mg/dL) and low levels of 1.25 $(OH)_2D_3$ (12 pg/mL, normal values 15–60 pg/mL).

Which is the most probable diagnosis?

a. Hyperparathyroidism
b. Vitamin D deficiency
c. Tumor-induced osteomalacia
d. Hypophosphatasia
e. Fanconi syndrome

Comment

A CT scan showed a lesion in the right thigh. Furthermore, an octreotide labeled scintigram showed intense focal update in the same region. Fractional phosphate excretion was 25% indicating inappropriate phosphaturia, while serum levels of potassium, urate and bicarbonate were within normal limits and urinalysis did not show glucosuria; thus, the diagnosis of Fanconi syndrome can be excluded. The levels of serum alkaline phosphatase were not low and this finding rules out hypophosphatasia, while the normal serum Ca^{2+} and PTH levels exclude the diagnosis of primary hyperparathyroidism. The absence of increased PTH levels can also exclude the possibility of vitamin D deficiency. The serum level of the phosphaturic hormone fibroblast growth factor (FGF-23) was markedly elevated. Thus, the patient exhibited a mesenchymal tumor-induced osteomalacia. In this disorder phosphatonines inhibit both the renal tubular absorption of phosphate and 1a- hydroxylase enzyme that converts 15(OH)D_2 to 1,25 $(OH)_2D_3$.

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Diagnosis: *Tumor-induced osteomalacia*