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Tumour-induced osteomalacia

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Introduction

Phosphate is important for normal mineralisation of bone. Phosphate is important in its own right for neuromuscular function, and profound hypophosphataemia can be accompanied by encephalopathy, muscle weakness and cardiomyopathy. Hypophosphataemia can be due to intracellular uptake of phosphate from the extracellular fluid, reduced intestinal phosphate absorption, increased renal excretion, decreased renal tubular absorptive capacity and genetic defects in renal tubule phosphate transporters.

Case presentation

A 31-year-old woman presented with hypophosphataemia (0.62–0.66 mmol/L), an abnormality she had since 2012. Fibroblast growth factor-23 (FGF-23) was above the upper limit of normal (98 pg/mL) but parathyroid hormone, calcium, full blood count, renal function test, random blood sugar, vitamin D and liver function test were normal. Her medical history included asthma, previous Ewing sarcoma in the chest wall for which she received chemotherapy, adjuvant radiotherapy and multiple chest wall operations in 2011. She was an ex-smoker. Family history was unremarkable. Based on history, examination and investigation, tumour-induced osteomalacia was the most likely diagnosis.

Discussion

FGF-23 plays an important role in the development of hypophosphataemic disease, such as tumour-induced osteomalacia, X-linked hypophosphataemic rickets/osteomalacia (XLH). It reduces serum phosphate by supressing proximal tubular phosphate reabsorption and intestinal phosphate absorption.

TIO is a rare cause of impaired bone mineralisation. Removal of the tumour resulted in rapid reduction in serum FGF 23 levels. In our patient with tumour-induced osteomalacia and XLH, FGF23 was above the upper limit of the reference range in most patients irrespective of medical treatment.

Phosphate and active vitamin D can be used in excessive action of FGF23 including a patient with TIO with unresectable tumours, but it has limited effects and several adverse events. Burosumab can be used in XLH. ■

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References

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