Hypercalcaemia aggravated: case report

A 61-year-old man developed aggravated hypercalcaemia while receiving vitamin-D.

The man was admitted due to weight loss of 10kg and acute renal failure. He has been receiving vitamin D 800 UI/day [dosage and indication not stated] for a few months. On admission lab test results were as follows: calcium 4.2 mmol/L, parathyroid hormone (PTH) 5 ng/mL, parathyroid hormone-related protein (PTHrP) <20 pg/mL, 25-hydroxy-vitD3 (cholecalciferol) 76 ng/mL, phosphate (PO4) 1.1 mmol/L, angiotensin-converting enzyme (ACE) <25, normocalciuria and no renal lithiasis. On osteodensitometry, lumbar osteoporosis and severe femoral osteopenia were noted. The whole body PET scan excluded a neoplastic lesion. Idiopathic interstitial pulmonary fibrosis was also detected. Lymph node samples rule out sarcoidosis. His hypercalcaemia aggravated [duration of treatment to reaction onset not stated].

Treatment with hydration, calcitonin, corticosteroids and zoledronic acid did not normalize serum calcium levels. Investigation showed 1,25-dihydroxyvitD3 (calcitriol) 161 ng/mL and Fibroblast growth factor 23 (FGF) 284 pg/mL, which suggested a functional deficit of CYP24A1 (enzyme responsible for the degradation of cholecalciferol and calcitriol). A genetic analysis by NSG confirmed a heterozygous mutation of CYP24A1 combining: c.62del (p.Pro21ArFs 8) and c.428.430 del (p.Glu143del). After administration of rifampicin for one year, calcium profile normalized. Test showed calcium 2.6 mmol/L, PTH 15 ng/mL and 1-25 vitD 46 pg/mL.

Parette MC, et al. Hypercalcemie aggravée par l’administration de vitamine D chez un patient adulte révélant une mutation hétérozygote composite CYP24A1 : réponse et traitement à long terme par rifampicine. Annales d’Endocrinologie 82: (plus poster) abstr. PA-088, No. 5, Oct 2021. Available from: URL: http://doi.org/10.1016/j.annder.2021.08.157 [French; summarised from an English abstract]