

Clinical case number 1

A 48-year-old non-smoker male patient presented with progressively worsening skeletal pain for the previous five years. X-rays of the lumbosacral spine and hip showed markedly reduced bone density with prominent trabecular markings. Dual-energy X-ray absorptiometry (DEXA) confirmed the presence of severe osteoporosis (T score <-2.5 in the lumbar vertebrae and left femoral neck). He also had impaired renal function, with serum creatinine concentration of 250 $\mu\text{mol/L}$ (reference interval 53–106 $\mu\text{mol/L}$) and serum urea concentration of 10 mmol/L (reference interval 2.5–7.5 mmol/L). He was referred to our hospital for further evaluation of his impaired renal function. The patient was not diabetic or hypertensive nor did he have any history of corticosteroid therapy. Full blood count showed mild normochromic normocytic anaemia: haemoglobin 10.9 g/dL (reference interval 12.0–15.0 g/dL). Total and differential white blood cell counts were within reference limits. Biochemical investigations showed hypophosphataemia (0.6 mmol/L , reference interval 0.8–1.5 mmol/L), hypouricaemia (100 $\mu\text{mol/L}$, reference interval 208–416 $\mu\text{mol/L}$) and increased alkaline phosphatase (412 U/L , reference interval 108–306 U/L). Other liver function tests were within the reference limits, as were the serum concentrations of sodium, potassium, calcium and magnesium.

Routine urinalysis showed trace albumin and 2+ glucose on urine dipstick but plasma glucose was normal. Fractional excretion of uric acid (18%, reference interval 5–11%) and phosphate (31%, reference interval 5–20%) were elevated. Urine protein:creatinine ratio was 310 mg/mmol (reference interval <22 mg/mmol), and urine albumin:creatinine ratio was 5.3 mg/mmol (reference interval <2.5 mg/mmol of creatinine). Subsequently, the patient was re-evaluated. On further questioning, the patient gave an occupational history of working in a silver jewellery industry for the past 15 years.

What is the right diagnosis?

What will be the treatment?