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CLINICAL QUIZ

A rare cause of hypercalcemia: Questions

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Case summary

A previously well 12-year-old boy presented with a 1-month history of polydipsia, polyuria, and lethargy. Over that period, he had been drinking at least 3 l of water daily, reported feeling thirsty, and needed to pass urine approximately every 30 min. His parents also reported that he had weight loss over the previous month with reduced appetite secondary to nausea. Of note, he had a long-standing history of drinking approximately 1 l of cow's milk daily. He had no recent acute illnesses or fevers and reported no pain, discomfort, or respiratory distress. He had been treated with azathioprine for 3 years in the past for intractable eczema. His blood glucose level checked by his general practitioner was normal.

The answers to these questions can be found at http://dx.doi.org/10.1007/s00467-013-2707-1

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Physical examination revealed significant bilateral inguinal lymphadenopathy and a 2-cm palpable liver edge. There were patches of dry skin attributed to previously diagnosed eczema. His cardiovascular, respiratory, neurological, ENT, and musculoskeletal examinations were otherwise unremarkable. There was an evident BCG scar. Vital signs were within normal limits.

Laboratory investigations revealed acute renal impairment, hypercalcemia, and a mild transaminitis: urea 15.2 mmol/l, creatinine 149 mmol/l, serum calcium 3.38 mmol/l, ionized calcium 1.78 mmol/l, serum phosphate 1.42 mmol/l, sodium 139 mmol/l, potassium 3.7 mmol/l, AST 76 U/l, ALT 114 U/l, and lactate dehydrogenase 416 U/l. Serum intact parathyroid hormone levels were suppressed at <6 ng/l. Serum 25(OH)-cholecalciferol level was reduced at 38 nmol/l. Urinalysis revealed significant hypercalciuria (calcium/creatinine ratio of 2.91). Full blood count measurements were within normal limits, while blood film revealed only occasional atypical lymphocytes and monocytes. An abdominal ultrasound revealed bilateral hyperechogenic kidneys, which were otherwise unremarkable; and mild hepatosplenomegaly with bulky inguinal lymph nodes bilaterally with speckled hyperechogenicity. A chest radiograph revealed clear lung fields, normal-sized cardiac silhouette with no evidence of a widened mediastinum.

Questions

1) What differential diagnoses would you consider to be possible causes of the hypercalcemia?
2) What is the most likely cause of his presentation?
3) What further diagnostic investigations would you consider?
4) How would you treat hypercalcaemia?