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LETTER TO EDITOR

Anemia in patient with primary hyperoxaluria and bone marrow involvement by oxalate crystals

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Abstract
We present a rare case of anaemia secondary to bone marrow infiltration by oxalate crystals and renal failure in a patient diagnosed with primary hyperoxaluria. In our case, the anaemia was recovered after the double liver and kidney transplantation, the latter was performed on two occasions after the failure of the first graft.

To the editor,

Primary hyperoxaluria is a rare autosomal recessive genetic disorder. It is caused by a deficiency of hepatic enzymes, which causes increased production of oxalic acid leading to hyperoxalemia and deposition of calcium oxalate crystals in the kidneys where nephrocalcinosis and nephrolithiasis develop leading to chronic renal failure. As a result of decreased glomerular filtration, oxalate is subsequently deposited in other tissues, particularly in the skeleton, causing systemic condition called oxalosis.

Three different types of primary hyperoxaluria (types 1, 2, and 3) have been identified [1], caused by different enzymatic deficiencies and the involvement of different intracellular organelles. Primary hyperoxaluria type 1, the most common type, has an incidence rate of approximately one case per 120,000 live births per year in Europe [2]. It is caused by a deficiency of the liver-specific peroxisomal enzyme alanineglyoxylate aminotransferase (AGT), which determines an increase in the urinary excretion of both oxalate and glycolate [1]. Without treatment, the prognosis of these patients is poor and combined hepatic–renal transplant is necessary for cure [3].

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Case report

A 41-year-old man with end-stage renal disease, secondary to nephrolithiasis and nephrocalcinosis, referred to the hematology clinic for the investigation of anemia with lack of response to erythropoietin. His past medical history was significant for renal colic and nephrolithiasis since the age of 18 years. Mild renal failure was diagnosed for the first time at the age of 35 years, and for the past 2 years he was on dialysis. There is no known consanguinity between his parents. Physical examination was unremarkable, without evidence of hepatosplenomegaly. Laboratory studies showed hemoglobin level of 8.7 g/dL, leukocyte count of 3.700/mm^3, platelet count of 219.000/mm^3, reticulocytes 3.1%, and frequent teardrop cells in the blood film. Computed tomography scan confirmed nephrocalcinosis and absence of hepatosplenomegaly, also it revealed generalized hyperdensity of the bones (Fig. 1). The bone marrow aspirate was dry tap. Bone marrow biopsy showed intertrabecular spaces occupied by abundant calcium oxalate crystals (Figs. 2 and 3), areas of fibrosis with fibroblastic proliferation and irregular trabecular bone remodeling (Fig. 4). Mutation analysis showed homozygosity for the mutation in AGxT (c.33dupC). Based on the previous findings a diagnosis of primary hyperoxaluria type I was made.

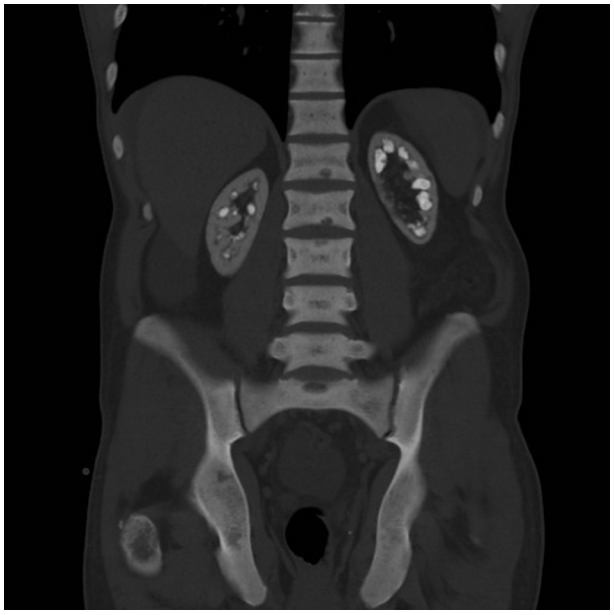

Fig. 1 CT scan abdomen and pelvis coronal view.

At the age of 45 years, the patient underwent double liver and kidney transplantation from a deceased donor. The renal graft never functioned and the patient continued dialysis. The liver graft is fully functioning with almost normal LFTs (Liver function tests). Hemoglobin level recovered to 11—12 g/L after liver transplantation and treatment with darbepoetin alfa 150 mcg/week.

At the age of 49 years, the patient underwent his second kidney transplant from a deceased donor during which he developed multiple complications. Currently his renal graft is functioning, maintaining serum creatinine level of 3.6 mg/dL at his last follow up, and dialysis was discontinued 1 month after the transplantation. Anemia was completely resolved, showing Hb of 15.2 g/L at his last evaluation, and the dose of darbepoetin alfa was reduced to 40 mcg/week.

A patient with a history of renal lithiasis, crystals of calcium oxalate in urine, and bone marrow led us to conclude that he presented a picture of systemic oxalosis, which should be conditioned by primary hyperoxaluria. Bone is the most common location of the deposit [1], however, to our knowledge, the only similar case of reversal pancytope-

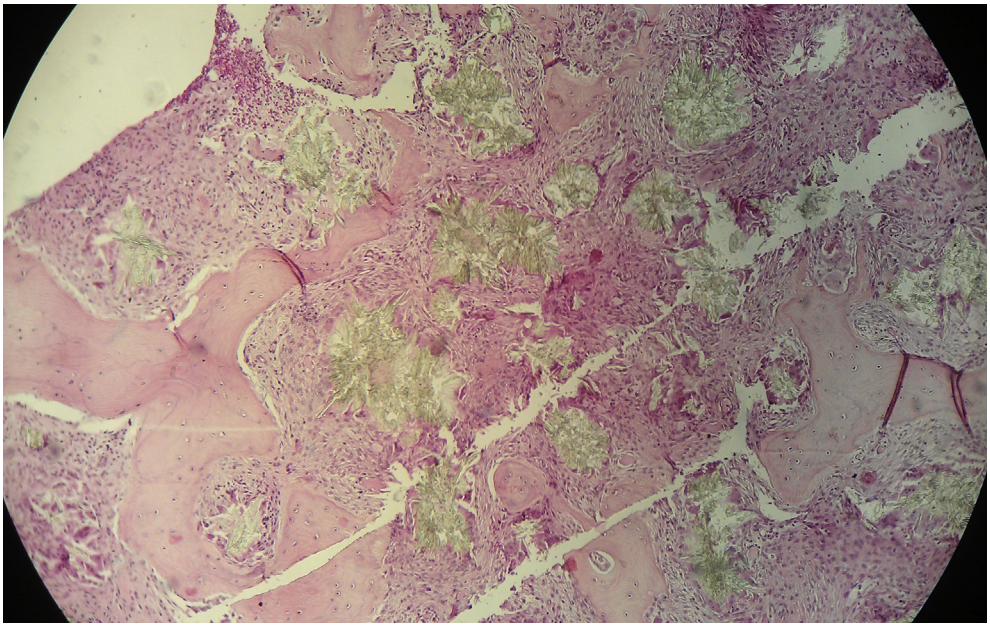

Fig. 2 Hematoxylin and eosin stain of the bone marrow, 20-fold magnification.

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