2,8-dihydroxyadenine urolithiasis: A case report

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The 2,8-dihydroxyadenine (2,8-DHA) lithiasis is a rare cause of urolithiasis, induced by a deficiency of the APRT (adenine phosphoribosyltransferase) enzyme. Early diagnosis is critical for this rare genetically-determined metabolic abnormality. Clinical manifestations of this disease are induced by a very low solubility of 2,8-DHA which crystallizes in the nephrons. First case of 2,8-DHA lithiasis described in Tunisia, in a 5-year old boy displaying a renal colic while systemic examination revealed no abnormality is reported. Abdominal ultrasound revealed the presence of calculi in the lower right cup and lower end of the right ureter. He received one session of extracorporeal shock wave lithotripsy. Fragments removed were analyzed by infrared spectroscopy in the laboratory of biochemistry in Charles Nicolle Hospital and objectified stones entirely consisting in 2,8-DHA. Confirmation of the diagnosis was made by determination of the APRT activity in erythrocytes by radiolabeling methods in the metabolic biochemistry laboratory of the Necker Hospital (Paris). This assay has objectified a total APRT deficiency. A genetic study, realized in the latter laboratory, has shown that enzyme deficiency is due to a newly identified gene mutation, never described so far. The child was placed under allopurinol dose of 10 mg/kg/day, associated with a diet low in purines and diuresis. Three years post-treatment, clinically the patient is stone-free while biologically, he kept preserved renal function. 2,8-dihydroxyadenine lithiasis is a severe stone disease whose frequency is under-estimated. Early diagnosis is essential to prevent formation of recurrent stones and occurrence of renal failure.

Biography

O Marrakchi has completed her MD, specialization degree and Assistant Professorship in Biochemistry at the Medicine Faculty of Tunis (Tunisia). She has obtained her University Diploma “Urinary Lithiasis” in 2010 at the University Paris-Sud (France), and specialized in biological exploration of urolithiasis through several trainings at the Necker Hospital in Paris. She has served as academic university-hospital assistant in Charles Nicolle Hospital of Tunis and was a member of the research unit “Urolithiasis” and has publications in this topic. She is presently directing a biomedical laboratory with specialization in the exploration of urinary stones.

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