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Late Revelation of Primary Hyperoxaluria with Renal, Ophtalmic and Hematologic Localization: Hyperoxalurie Primitive De Révélation Tardive Avec Localisation Rénale, Ophtalmologique et Hématologique

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Abstract **Case Report**

Primary hyperoxaluria is a rare heredetary disease, with autosomal recessive inhertance. It's metabolic abnormality with responsible for accumulation of calcium oxalate deposits. We report the case of a young patient, 22 years old, born of first consanguineous marriage. He is at end chronic renal disease of unknowm nephropathy. This patient has been in hemodialysis for 4 years. He presented with anemia resistant to erytropeitin associated to bone pain. The diagnosis of hyperoxaluria was suspected in the presence of urolithiais and confirmed by the presence of both ophthalmic and bone marrow calium deposit. The therapy consisted in iterative transfusions, vitamintherapy with daily hemodialysis. The follow up was stable. Waiting for combined kidney and liver transplantation.

Keywords: Primitive hyperoxaluria, oxalate deposit, resistance to erytropeitin.

INTRODUCTION

Oxalate is the ionic form of oxalic acid, metabolized in the liver and excreted in the urine; primary hyperoxaluria is a rare genetic abnormality, characterized by excess endogenous oxalate production, and therefore increased urinary excretion. Oxalosis brings symptoms arising from deposits in different tissues together. It is a rare disease, of often late revelation with fatal consequences.

CASE REPORT

We report the case of a 22-year-old Moroccan black patient who was referred for an etiological evaluation of bone pain with anemia erythropoietin resistance in our exercise. This patient is the youngest of five siblings, three of whom died from unknown cause in childhood; he is born from a marriage between blood relations of 1st degree. The history of his kidney disease seems to go back to 4 years, by the discovery of chronic renal failure at the terminal stage, on indeterminate nephropathy, revealed by a uremic syndrome, the patient was initiated on hemodialysis on femoral catheter then on fistula arterioveinous native. Moreover, the patient does not disclose any other related extrarenal signs or history of recurrent urinary tract infections, nor of stone emission and without notion of familially recognized lithiasic pathology.

The onset of current symptoms dates back to one year, with bone pain settling that gradually increases in severity, associated with normocytic normochromic anemia resistant to maximum doses of erythropoietin. On clinical examination: the patient was conscious, black skin, normo stretched to 130 / 80mmhg, with a skin-mucous pallor. Left aneurysmal radial arteriovenous fistula with a good thrill, the residual The residual diuresis was null, with no bone pain points or joint deformities or stiffness or skin lesions. Tumor syndrome was not reported on the abdominal and lymph node tests. The remainder of the somatic examination was unremarkable.

Biologically, the hemogram objectived an anegenerative normo chromium normocytic chromium anemia at 6.6g / dl, with a ferritinemia at 1188 ug / l, a serum iron at 0.77ug / dl; transferrin et 1.4g / l, a mild inflammatory syndrome was found on plasma protein electrophoresis, with a negative CRP and VS respectivly, and a b2 macroglobulin at 35.14. Hemoglobin electrophoresis was normal.

The proportion of serumal aluminium had not objectified intoxication (aliminium level was =15). The rest of the biological sheeck up had shown a parathormone in 328, the proportion of the vitamin D with a phosphocalcique normal balance sheet. Blood smear was normal, as well as the rate of vitamin B12.

On the radiological evaluation, the plain urinary tract showed multiple symmetrical bilateral calcium images on the kidney projection (Figure-1), with the ultrasound counterpart, an illustration of common progressive nephrocalcinosis: a shadow cone on the renal compartments limiting the individualisation of the kidneys. However, no deep lymphadenopathy with abdominal ultrasound, The rest of the radiological examination of the skeleton revealed no abnormalities.

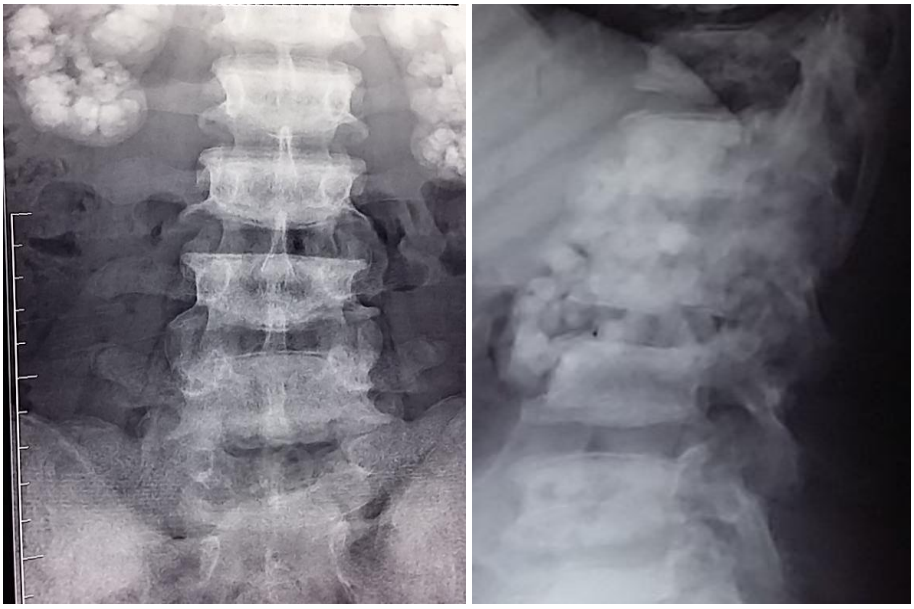

Fig-1: Plain urinary tract demonstrated multiple symmetrical bilateral calcium images on the kidney projection

The diagnosis of primary hyperoxaluria was strongly suspected, with probable involvement of the bone marrow explaining EPO resistant anemia. An osteomedullary biopsy was performed with spinal aspiration confirming our hypothesis, with the presence of deposits of calcium oxalate crystals, associated with spinal fibrosis and a macrophagic reaction (Figure-2).

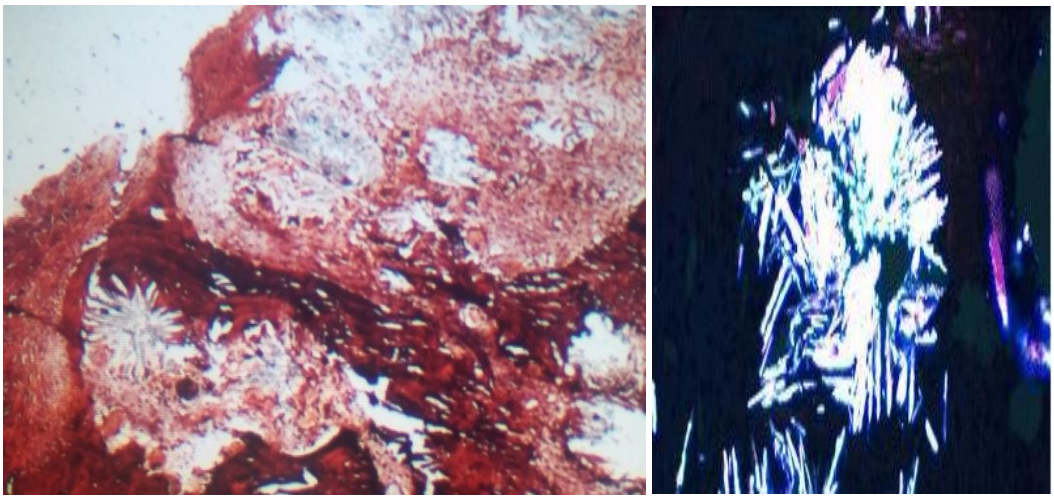

Fig-2: osteomedullary biopsy demonstred the presence of deposits of calcium oxalate crystals, associated with spinal fibrosis and a macrophagic reaction, with birifringence in polarized light

The diagnosis of primary hyperoxaluria I was be confirmed. The extension cheekup of this disease was accomplished, in particular an ophthalmic exploration putting in abvious retinal stores at the funduscopic examination (Figure-3), with normal visual acuity. Cardiac exploration did not show any abnormalities, with good cardiac function, (LVE at 53%), without images of calcifications or electrocardiogram disturbances.

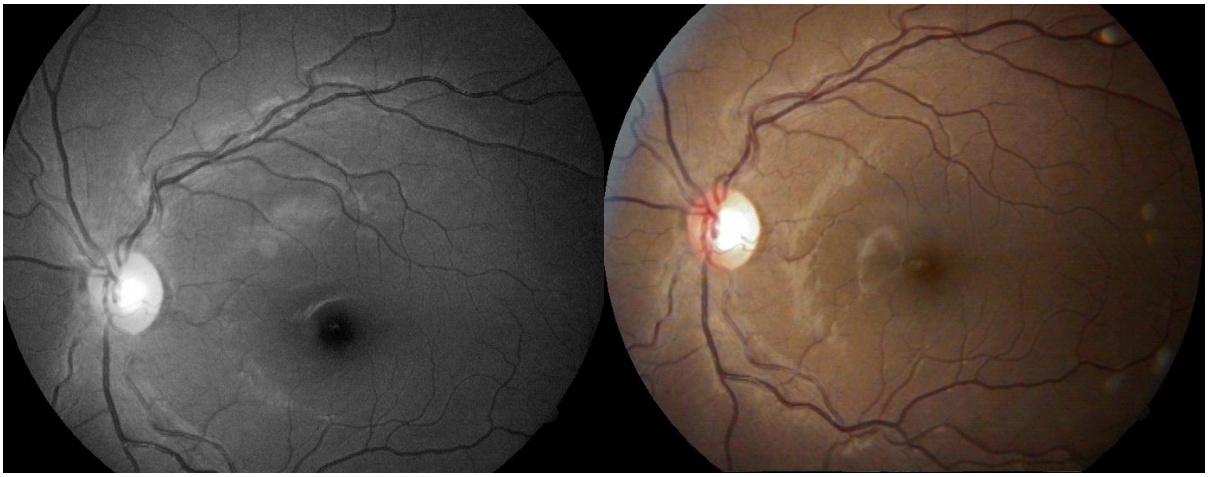

Fig-3: Some retinal stores at the funduscopic examination

Pending bouth kidney and liver tranplantation A regular monitoring was planified, the treatement was based on vitaminotherapy such as pyridoxine, intensified dialysis and iterative transfusion if necessary.

DISCUSSION

Primary hyperoxaluria is a congenital metabolic abnormality caused by mutation of the AGXT gene (alanine glyoxylate aminotransferase), an enzyme that is formed only in the liver and whose principal coenzyme is pyridoxine phosphate (vitamin B6). Its deficit is responsible for the uncontrolled production of insoluble calcium oxalate and thus induces its deposition in the different organs and tissues [1-5].

It is a rare and often ignored pathology, its estimated incidence in France is of the order of one birth in 120,000. Primary hyperoxaluria I presents 0.5% of the causes of chronic end-stage renal disease in children in Europe [4, 6, 7].

The clinical symptomatology is diverse, secondary to calcium oxalate deposits in the various tissues. The renal disease progresses towards the terminal stage in a variable manner, it is due to the inflammation and tubular obstruction attributing to a fibrosis and leading to the degradation of the renal function. From a renal function of 30ml / min / m2 of GFR, the excretion of oxalate deteriorates with increased possibility of these deposits at the tissue level [8].

Other manifestations are possible and should be looked for systematically, ophthalmic involvement marked by retinal deposits of calcium oxalate that appear in founduscopic examination, as was the case of our patient. Cardiac involvement such as valvular calcifications, rhythm disturbances or myocardial ischemia. Bone involvement, with anemia resistant to erythropoietin by central involvement [8, 9].

Other damage is likely but remains rare, vascular, neuromuscular and skin involvement [1, 4].

The diagnosis of the disease is often late, at the stage of complications, which makes its prognosis even mediocre and difficult to manage. It is based on the combination of clinical, biological and genetic arguments [2]. On a family history suspected in the anamnesis, the radiological examinations are verry interesting, they showed renal stones [2, 10] whose morphological analysis by infrared spectrometry determines their nature. One of the biological tests for diagnosis is urine oxalate on a 24-hour urine, or the urine glycolate and glycerate assay. In the event of a chronic renal failure due to oligo-anuria, these dosages remain difficult and uninterpretable [2, 10]. The high blood oxalate dosage or the oxalate to creatinine ratio in plasma can therefore, particularly in this entity, contribute to the diagnosis (value greater than 80umol / l). Other assessments can help confirm the diagnosis of primary hyperoxaluria, it is the demonstration of specific genetic abnormality, as well as hislogical déposits of calcium oxalate in organ [2, 4, 11-13].

Therapeutically, Hyperhydration is recommended, it allows to increase the solubility of calcium oxalate but of negligible impact and remains contraindicated at the IRCT stage [1, 9, 10]. Citrates are also recommended as crystallization inhibitors, but are prescribed with great caution when renal function deteriorates [1, 14].

Vitamin B 6 or oral pyridoxine remains the treatment considered useful in end-stage renal disease, a reduction of almost 30% of excreted oxalate was noted [1, 14].

Probiotics which include O. Formigen, which uses oxalate as a source of energy and thus reduces blood levels, was shown to be effective in animals but not yet tested in humans [4, 15].

Kidney transplantation alone has a high probability of graft loss due to recurrent deposits. The treatment of choice remains bouth kidney and liver transplantation [4, 14, 15].

Screening for the disease is essential in order to improve their prognosis in front of any child with a family history with an episode of renal stones [16, 17].

CONCLUSION

Through this observation, we raise the point on the early diagnosis of this rare disease before the stage of complications which can involve the functional and vital prognosis of the patient.

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