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PRIMARY HYPEROXALURIA IN MACEDONIAN CHILDREN

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Primary hyperoxalurias (PH) are rare metabolic disorders due to liver deficiency of specific enzymes leading to recurrent nephrolithiasis, nephrocalcinosis and renal failure. We present two children with PH who were diagnosed late. Case 1: Twelve-year old boy with gross hematuria, colicky pain and massive bilateral nephrolithiasis. Rejection of surgical treatment and poor compliance with conservative measures resulted in prompt progression to terminal renal failure manifesting severe complication of the disease (EPO-resistant anemia, oxalate retinopathy, osteopathy, cardiomyopathy). Diagnosis of PH-1 was based on elevated urinary values of glycolate. The boy had HBV and HCV coinfection and died due to liver cirrhosis. The second patient displayed hematuria and solitary calculus in the right kidney at the age of four years. Urinary oxalate was not measured at that time. The calculus was successfully extracted. At the age of twelve the ultrasound scan and excretory urography revealed bilateral massive nephrolithiasis. Urinary oxalate was elevated (1,80 mmol/1,73m2/d). Glycolate and L-glycerate were within referent values. Based on these results the type of PH could not be determined. He showed partial response to high pyridoxine dose. The boy underwent classical surgery to relieve the obstruction. His GFR is mildly impaired but stable under aggressive conservative treatment. In conclusion: oxalate measurement is mandatory in all children with nephrolithiais, early diagnosis and aggressive treatment may prevent complications of the disease.