Bilateral Renal Stones Associated with Nail-Patella Syndrome

R.	Ravindra	Mittal
S.	Sanjiv	Saxena
R.K.		Hotchandani
S.K.		Agarwal
S.C.		Tiwari
S.C.		Dash

Department of Nephrology, AIIMS, New Delhi, India

Dr. Sanjiv Saxena, Assistant Professor, Department of Nephrology, AIIMS, New Delhi-110029 (India)

Dear Sir,

The nail-patella syndrome (hereditary osteo-onychodysplasia or HOOD) is a well-recognized autosomal dominant disorder characterized by nail, skeletal and renal abnormalities. A spectrum of renal involvement has been described in patients with the nail-patella syndrome [1,2].

Although Glassock et al. [3] mention that a disproportionate number of these patients may have renal stones (references not quoted), on review of the literature, we came across only three case reports describing such an association [2, 4, 5]. However, all the above three cases had evidence of unilateral calculus disease.

A 23-year-old male was referred to the Nephrology Services of All India Institute of Medical Sciences, New Delhi in June 1993 for the management of chronic renal failure. He had a history of recurrent bilateral renal colics for the preceding 3 years and had undergone pyelolithotomy on the right side and ureterolithotomy on the left side for bilateral calculus disease 2 years earlier in a district hospital. The status of his renal functions at the time of surgery was not known. The patient had no subsequent colics and repeated skiagrams revealed no radiopaque calculi. However, for the preceding 4 months, he started having anasarca and was found to have mild hypertension and moderately severe azotemia. There was no history of renal calculus disease or any other renal illness in the family.

Clinical examination revealed severe pallor, mild hypertension and changes of chronic azotemia. In addition, the patient had dystrophic nails in both upper and lower limbs, fixed flexion deformity of the right elbow, bilaterally small hypoplastic patellae and bilaterally palpable iliac horns. There were surgical scars of pyelolithotomy and ureterolithotomy on the right side and left side of the abdomen, respectively. Neither of the kidneys was palpable.

Laboratory evaluation confirmed the presence of severe anemia with Hb 0.70 mmol/l (4.7g/dl) and severe azotemia with serum creatinine of 1,408 μ mol/l (16.0 mg/dl). Urine examination revealed 2+ proteinuria and 8-10 WBCs per high power field on microscopy. 24-hour urine examination showed 1.98g proteinuria and a creatinine clearance of 4 ml/min. Skeletal survey revealed bilateral iliac horns, bilaterally hypoplastic patellae and an ill-developed capitulum of right radius. Ultra-sonography of the abdomen revealed bilateral normal-sized kidneys with a

grossly irregular surface and mild dilatation of pelvi calyceal system on both sides. The cortices were bilaterally thinned out. No calculus was seen presently on either plain skiagraphy of the abdomen or on ultrasonography.

A clinical diagnosis of nail-patella syndrome with operated bilateral renal calculus disease and end-stage renal failure was made in view of the typical nail and skeletal changes present in the patient. The patient was subsequently put on biweekly maintenance hemodialysis and was proposed for renal transplantation.

The nephropathy in the nail-patella syndrome is indeed quite varied. For unknown reasons, renal disease is not always present in patients with the nail-patella syndrome (exact percentage not determined). Looij et al. [1] reported that renal disease occurred on approximately 48% of the subjects with osteo-onychodysplasia. It can be asymptomatic with moderate proteinuria for many years, or associated with microhematuria. Bennett et al. [2] had earlier reported that more than half of the patients with skeletal abnormalities had an abnormal urinary sediment, impaired urinary concentrating ability or abnormalities in urinary acidification or urinary protein excretion although the nephrotic syndrome is rare in these patients. Progression to end-stage renal failure occurs in a variable number of patients, ranging from 10 to 30% in different series [1,2]. A significant proportion of these patients were reported to have congenital urinary tract malformations [2, 3]. Glasscock et al. [3] also mentioned that renal stones occurred in a disproportionate number of patients with the nail-patella syndrome; however, no references have been quoted. On review of the literature, we came across only three case reports describing the presence of renal calculus in patients with the nail-patella syndrome [2,4, 5]. In all these three cases there was evidence of only unilateral calculus disease. Our case is the first case report describing the presence of bilateral renal calculus disease in a patient with the nail-patella syndrome.

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