Ehlers-Danlos Syndrome with Reflux Nephropathy

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Dear Sir,

Ehlers-Danlos syndrome (EDS) is a disorder of connective tissue with cutaneous, skeletal, ocular and visceral manifestations. Urinary tract abnormalities including uretero-pelvic anomalies [1], medullary cystic disease of the kidneys with renal tubular acidosis [2], bladder diverticuli [3], and hypoplastic kidney [3] have been infrequently reported in EDS. We describe a case of EDS with reflux nephropathy.

A 30-year-old male was admitted in April 1994 with end-stage renal failure and hypertension. In October 1992, during an ultrasound evaluation undertaken to determine the cause of a right upper quadrant pain, he was detected to have a right kidney measuring 11 cm with moderate dilatation of the pelvicalyceal system. The left kidney measured 6.9 cm. He also had gallstones. Investigations revealed a serum creatinine of 2 mg/dl with 2+ proteinuria. The 24-hour urine estimation for protein was 1.07 g. A MAG-3 renogram revealed no uptake on the left side and slow uptake of the radiopharmaceutical excretion on the right kidney with delayed excretion. Voiding cystourethrogram revealed a grade 4 vesicoureteric reflux on the right side with tortuosity of the ureter. There was a larger bladder diverticulum arising from the left posterior wall. His past medical history included severe laxity of his joints including the small joints of his hands with recurrent subluxations and dislocations. He had severe kyphosis and underwent surgery with anterior and posterior spinal fusion with Harrington rod placement in 1983. He also had an inguinal hernia and mild mitral regurgitation. His family history was negative for EDS, urinary infections and renal failure.

On examination his pulse was 80/min, blood pressure 160/94 mm Hg. His height was 1.65 m and he weighed 40 kg. He had a redundant skin and there were multiple paper-thin scars over his forehead. He had kyphoscoliosis. There were spontaneous dislocation of his proximal interphalangeal, distal interphalangeal and metacarpophalangeal joints. The knee and elbow joints were hyperextensible. Cardiac auscultation revealed 2/6 systolic murmur. Examination of other systems was unremarkable.

Investigation revealed a serum potassium of 7.5 mmol/l, serum bicarbonate of 16 mmol/l, serum chloride of 94 mmol/l, serum creatinine of 8.6 mg/dl and blood urea nitrogen of 81 mg/dl. Hemoglobin was 14.1 g/dl and platelet count was $299 \times 10^9/l$. He was dialyzed emergently by a double lumen subclavian Quinton catheter and his condition stabilized after three hemodialyses. An arteriovenous fistula was constructed on his left arm for chronic dialysis. He remains stable on dialysis and is being evaluated for a renal transplant.

Zalis and Roberts [3] reported hypoplastic kidneys and bladder diverticulum in EDS, however VCUG was not done in their case. Bladder diverticulum has been reported in EDS type 9 associated with bladder neck obstruction and occipital horns [4]. We believe that this is the first case of reflux nephropathy reported in EDS. Earlier reports have suggested laxity of the bladder wall as a cause of bladder diverticuli in EDS [3]. We believe that in the absence of a blad-

der neck obstruction, laxity of the connective tissue resulted in incompetence of the vesicoureteric junction and the diverticuli in our patient. The small left kidney could be a congenital hypoplastic kidney, however in the absence of an angiogram we cannot exclude the possibility of ischemia from renal artery stenosis. Renal transplantation remains an Achilles heel given the overall nature of this disease.

References

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