Steinstrasse due to distal renal tubular acidosis with sensorineural deafness

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Keywords: distal renal tubular acidosis, hypercalciuria; nerve deafness; nephrocalcinosis; Steinstrasse

Introduction

Primary distal renal tubular acidosis (DRTA) is characterized by metabolic acidosis of varying severity accompanied by an inappropriately alkaline urine. Other features include low serum potassium due to renal potassium wasting, and elevated urinary calcium. If untreated, this acidosis may result in dissolution of bone, leading to osteomalacia and rickets. Renal deposition of calcium salts (nephrocalcinosis) and renal stone formation commonly occur [1].

Primary DRTA is inherited as either an autosomal dominant or autosomal recessive trait [2–4]. Patients with recessive DRTA are severely affected, presenting with either acute illness or growth failure at a young age. In addition, hearing loss is confined to a subset of patients with recessive DRTA [4–6]. We report a patient with DRTA and sensorineural deafness who presented with nephrocalcinosis and developed spontaneous Steinstrasse affecting the right ureter.

Case

A 20-year-old man presented with failure to thrive and skeletal deformity (bilateral genu valgum). He had a history of vomiting and dehydration when a few months old. On the basis of growth failure, hyperchloaemic metabolic acidosis with alkaline urine and hypokalaemia, a diagnosis of DRTA was made. An audiogram showed moderate to severe bilateral sensorineural hearing deafness, and he was prescribed hearing aids. Bilateral nephrocalcinosis in a papillary distribution was demonstrated by abdominal X-rays.

His urinary calcium excretion ranged from 2.8 to 4.1 mg/kg per day. No glucosuria, hyperaminoaciduria or hyperphosphaturia were found. After diagnosis of DRTA with nerve deafness, the patient was maintained on oral treatment with sodium bicarbonate at a dose ranging from 72 to 96 mEq per day and potassium citrate 20–30 mEq per day and high fluid intake. The genu valgum was treated by corrective osteotomy. Impaired growth and progressive deformity were improved by sustained correction of metabolic acidosis with alkali supplementation.

After an 8-year follow-up period, the patient’s glomerular filtration rate remained stable, the nephrocalcinosis did not progress, and his height increased 10 cm. He passed occasional stones. The hearing loss was not modified. Further studies were performed when the patient was 28 years old. Urinary pH was 7.10, and titratable acid excretion and ammonium excretion were 19 and 8 μEq/min per 1.73 m² body surface, respectively, during spontaneous metabolic acidosis (blood pH 7.30, serum bicarbonate concentration 18 mEq/l). The fractional bicarbonate excretion was 4.6% in the presence of a normal serum bicarbonate concentration (22.8 mEq/l) induced by bicarbonate supplementation. The urine minus blood pCO₂ differences (U–B pCO₂) were equal to 0 mmHg, in alkaline urine (urine pH 7.24) following oral bicarbonate administration, and 5 mmHg after a sodium phosphate load (urine pH 7.16, urine phosphate concentration 17.5 mmol/l).

He was initially compliant with therapy. However, when the patient was 29 years old, due to personal problems, he became noncompliant with bicarbonate and citrate treatment. A plain abdominal film showed bilateral nephrocalcinosis in a papillary distribution and Steinstrasse involving the right ureter, extending from the ureteropelvic junction to the distal ureter (Figure 1).

Discussion

The syndrome of DRTA and sensorineural deafness is a distinct nosological entity that is inherited as an
The clinical presentation of Steinstrasse may be associated with renal colic or may be silent. Spontaneous large Steinstrasse (occupying more than a third of the ureteral length) is very rare. To our knowledge, massive spontaneously occurring Steinstrasse associated with DRTA has been described in only two cases [8,9]. However, this association has not been reported in DRTA with nerve deafness.

Nephrocalcinosis in our patient was first noted during his initial evaluation. Once oral alkali therapy was started, the progression of nephrocalcinosis ceased and the frequency of stone passage decreased. Therefore, in this patient the cause of worsening of nephrocalcinosis, intermittent stone passage and silent Steinstrasse formation was without doubt due to non-compliance with sodium bicarbonate and potassium citrate treatment.

References


Steinstrasse is mainly a complication of extracorpor-