Nephrocalcinosis in Shwachman's syndrome

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SUMMARY Nephrocalcinosis has been reported only infrequently in Shwachman's syndrome. We describe a case in which nephrocalcinosis occurred and speculate that this may be due to increased urinary oxalate excretion.

The syndrome of exocrine pancreatic insufficiency and bone marrow dysfunction was first described by Shwachman and colleagues in 1964, and subsequently other features such as bone dysplasia have been recognised.1 In 1980, Aggett et al reviewed a series of 21 patients, one of whom was found to have nephrocalcinosis at necropsy.2 We report another child with Shwachman's syndrome in whom nephrocalcinosis was an early feature.

Case report

A boy, weighing 3300 g, who was the second son of healthy, unrelated parents was born after a normal pregnancy, labour, and delivery. He was initially fed orally on a standard infant formula, but feeding became increasingly difficult and he failed to thrive. At 6 weeks of age, he was admitted to hospital, where he continued to gain weight poorly despite numerous feed changes.

Nasogastric feeding was therefore started at 10 weeks of age. Investigations showed intermittent neutropenia (the lowest concentration was <0-2×10⁹ neutrophils/l), persistently low serum immuno-reactive trypsin (0-2 μg/l), and mildly raised plasma alanine transaminase and aspartate transaminase activities. Sweat sodium and chloride concentrations were normal. A radiograph of the chest showed broadened anterior ends of the ribs.

A diagnosis of Shwachman's syndrome was made and he was treated with pancreatic enzyme supplements from 4 months of age. Severe feeding difficulty and failure to thrive persisted, however, and at 7 months he was referred to us for further investigation. On admission his weight (5450 g), length (60 cm), and head circumference (42 cm) were all below the third centile. He had a narrow chest with Harrison's sulci, mild subcostal recession, increased tone in the legs with brisk reflexes and ankle clonus, and mild hirsutism. His liver was not enlarged. Pancreatic enzyme supplements were withheld on admission. Shwachman's syndrome was confirmed by low bicarbonate production (maximum 16 mmol/l), low lipase activity (maximum 13 μmol/min/ml), and absent trypsin activity in duodenal fluid obtained after stimulation with secretin and cholecystokinin, together with intermittent neutropenia and typical radiological changes in the ribs.

His transaminases were moderately raised (alanine transaminase 187 IU/l, and aspartate transaminase 122 IU/l). Immunological investigation showed poor neutrophil mobility. Further evaluation of the urinary tract was undertaken (see below) and pancreatic enzyme supplements were restarted at 8 months of age.

RENAL INVESTIGATIONS AND MANAGEMENT

Brightly reflective areas around the renal pyramids consistent with nephrocalcinosis had originally been observed in both kidneys on ultrasound examination at the age of 2½ months. At that time the urine was sterile and a spontaneous urinary pH of 5 was observed. Glycosuria or aminoaciduria were not present. The 24 hour urinary oxalate excretion was 0-31 mmol/1·73 m² surface area/24 hours (normal ≤0-46 mmol/1·73 m² surface area/24 hours). The 24 hour urinary calcium excretion was normal, as were the plasma urea and creatinine concentrations.

Further renal ultrasound examinations at the ages of 3 and 5½ months continued to show nephrocalcinosis, more noticeable on the left than on the right. At 7½ months, ultrasound examination showed nephrocalcinosis on the left side only; computed tomography of the abdomen performed at the same time confirmed the left sided nephrocalcinosis. Both investigations also showed dilatation of the right renal pelvis and ureter. A stone was visible in the right distal ureter on plain radiographs and computed tomograms.

The urine remained sterile, without aminoaciduria or glycosuria. Urinary calcium to creatinine ratio was 0-097 mmol/mmol (normal range 0-060-0-740 mmol/mmol), and oxalate excretion was 0-40 mmol/1·73 m² surface area/24 hours. At 8 months a right ureterolithotomy was performed. Analysis of the stone showed it to be 90% calcium oxalate.

At 11 months, after three months of continuous pancreatic supplementation, the urinary oxalate had
Nephrocalcinosis in Shwachman’s syndrome

We present a case of a 30 month old boy who aspirated a large amount of dirty sand and was successfully resuscitated and made a full recovery.

Successful treatment after ‘drowning’ in sand

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SUMMARY A 30 month old boy who aspirated a large amount of dirty sand was successfully resuscitated and made a full recovery.