

## Primary Hyperoxaluria

### Case History

An 18-year-old male presented with complaints of abdominal pain. He had history of previous episodes of renal colic and passage of stones in urine. Renal function tests were deranged (blood urea levels of 72 mg/dL and serum creatinine levels of 3.2mg/dL). Abdominal X-ray (Fig.1) and non-contrast computed tomography (NCCT) (Fig. 2) of the abdomen was performed.

### What is the Diagnosis?

Abdominal X-ray (Fig. 1) shows markedly increased density of both kidneys in the absence of contrast administration. Unenhanced CT abdomen (axial and coronal views) (Figs. 2a and 2b) reveals normal sized kidneys with symmetrical, markedly increased attenuation of the cortex of bilateral kidneys, compatible with cortical nephrocalcinosis. Small renal calculi were also seen (arrow showing left renal calculi) in both kidneys. The presence of diffuse symmetrical cortical nephrocalcinosis in normal sized kidneys, along with the history of passage of stones in urine, suggested a diagnosis of hyperoxaluria. The 24-hour urinary excretion of oxalic acid was markedly elevated, measuring 164 mg (normal 10 to 55 mg/24 h). Liver biopsy confirmed the diagnosis of primary hyperoxaluria.

### Discussion

The differential diagnosis of cortical nephrocalcinosis includes acute cortical necrosis, chronic glomerulonephritis, Alport syndrome, primary and secondary hyperoxaluria and rejected renal transplants.<sup>1-3</sup> Cortical nephrocalcinosis is quite uncommon and the medical history itself can give a clue to the underlying condition. The pathogenesis of acute cortical necrosis is multifactorial and usually follows a severe insult to the kidney, usually after placental abruption, septic abortion, transfusion reactions, burns, snake bite, severe dehydration and shock. These underlying risk factors were not present in the index case. Besides, the typical CT findings of acute cortical necrosis (enhancement of medulla, non-enhancement of renal cortex, thin rim of enhanced subcapsular tissue-the “rim sign”) were not seen in our case. Chronic glomerulonephritis occurs when there is slow, progressive destruction of the glomeruli of the kidney, with progressive loss of kidney function. Cortical



Fig. 1. Plain abdominal X-ray shows markedly increased density of both kidneys.



Fig. 2a. Unenhanced CT abdomen (axial view) reveals normal sized kidneys with symmetrical markedly increased attenuation of bilateral kidneys, compatible with nephrocalcinosis.



Fig. 2b. Coronal reformatted unenhanced CT abdomen reveals increased attenuation of bilateral kidneys, with small left renal calculi (arrow).

nephrocalcinosis due to chronic glomerulonephritis usually shows small kidneys, while the kidneys were normal sized in the index case. Alport syndrome is a hereditary nephritis, associated with nerve deafness, which was not present in our case. The presence of diffuse symmetrical cortical nephrocalcinosis in normal sized kidneys, along with the history of passage of stones in urine, suggested a diagnosis of hyperoxaluria.

Primary hyperoxaluria is a rare autosomal recessive metabolic genetic disorder, in which a defective glyoxalate metabolism in the liver, leads to increased oxalate production.<sup>1,2</sup> This results in abnormal deposition of calcium oxalate in bones, joints, nerves, heart, vessels, skin, retina and kidneys. There is also increased urinary excretion of oxalates and glycolate with subsequent formation of calcium oxalate stones and medullary (95%) or cortical (5%) nephrocalcinosis.<sup>2</sup> Secondary oxaluria can be due to impaired excretion, excessive oxalate intake, or increased absorption of oxalates in patients with chronic inflammatory bowel disease, small bowel resection, intestinal bypass and external biliary drainage.<sup>2</sup>

Primary hyperoxaluria can present in 3 forms: the neonatal and infantile forms, which are severe and include failure to thrive and rapidly progressive renal failure in early childhood and the more common, less severe, late childhood form (as in our index case) which usually present with renal calculi or nephrocalcinosis. Progressive renal failure in this form usually occurs in early adulthood. Calcium oxalate crystals are also deposited in extrarenal sites leading to extensive soft tissue and vascular calcification.

The diagnosis is established by demonstrating the increased levels of plasma oxalate, urinary glycolate and oxalate levels. Enzymatic study from liver biopsy can quantify the alanine-glyoxylate aminotransferase activity.<sup>1</sup> Initially the serum urea and creatinine levels may be normal and subsequently increase when renal failure sets in. Early detection and preventive treatment of the onset of renal compromise is important. Once renal failure sets in, surgical transplantation remains the only effective treatment, with combined liver-kidney transplantation being preferred.<sup>2</sup>

#### REFERENCES

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Niranjan Khandelwal,<sup>1</sup>MD, DipNB, FICR, Kushaljit Singh Sodhi,<sup>1</sup>MD, Vivek Virmani,<sup>1</sup>MD, DipNB, Navneet Sharma,<sup>2</sup>MD

<sup>1</sup> Department of Radiodiagnosis

<sup>2</sup> Department of Internal Medicine

Postgraduate Institute of Medical Education and Research, Chandigarh, India

Address for Correspondence: Dr Kushaljit Singh Sodhi, Department of Radiodiagnosis, PGIMER, Chandigarh, 160012 India.  
Email: sodhiks@gmail.com