A case of oxalate retinopathy in a patient with Primary Hyperoxaluria - a case report.

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Abstract: Mr. Murugan, a 30 year old male, the first born child of a non-consanguineous marriage. He was apparently normal until last August 2011 after which he suddenly developed breathlessness and vomiting. He was diagnosed to have severe renal failure (serum creatinine 10.2 mg/dl). CT scan of the abdomen showed bilateral nephrocalcinosis. Patient was diagnosed to have Renal Hyperoxaluria based on renal biopsy report, histopathological reports and calculus study. Ophthalmic evaluation showed whitish yellow deposits of oxalate crystals superior and inferior to the optic disc and palor of the Optic disc. Patient is awaiting a combined liver and kidney transplantation from a cadaver donor.

Keyword: Oxalate retinopathy, Primary Hyperoxaluria

INTRODUCTION: Primary Hyperoxaluria (PH) is a rare genetic (inherited) disorder that is present at birth. Primary hyperoxaluria comes in many forms, of which only two, Type I and Type II are well characterized. Type I and Type II are both autosomal recessive diseases. Although Type I is more common than Type II, it is still a rare disease with an estimated frequency between 1 in 100,000 and 1 in 1,000,000 1. It is a rare metabolic disorder caused by an enzyme defect which leads to an increased conversion of glyoxalate to poorly soluble oxalate which is then excreted in the urine 2. Most common renal complications include chronic renal failure, urolithiasis, renal colic, hematuria and acute renal failure due to complete obstruction 3. Wide spread deposits of oxalate in other organs leads to manifestations like cardiac conduction defects, hypertension, distal gangrene, reduced joint mobility and pain 4. Deposition of oxalate crystals in the retina and macula also occur 5. The main modality of treatment is a combined liver and kidney transplant.

Patient Details
Mr. Murugan, a 30 year old male, the first born child of a non-consanguineous marriage. He was apparently normal until last August 2011 after which he suddenly developed breathlessness and vomiting. He was diagnosed to have severe renal failure (serum creatinine 10.2 mg/dl). A CT scan of the abdomen was done which showed bilateral nephrocalcinosis.

Histopathology report
Patient was then sent for a Renal Biopsy. The reports were as follows:

- Fibrosis of the surrounding tissues.
- The universal tubules are atrophic.
- The crystals are identified as oxalate because of their birefringence under polarized light.
- More than 75% of the glomeruli are sclerotic and the remaining shows periglomerular fibrosis.
- The arterial walls are thickened and oxalate crystals are seen within them. The pelvis and ureter show no significant pathology.
Diffuse and global glomerulosclerosis with severe tubulointerstitial nephritis, severe arteriosclerosis associated with extensive birefringent crystal deposits - consistent with oxalosis. A calculus study was also done and revealed Calcium oxalate, non-calcium oxalate, carbonate-positive calculi.

**Diagnosis:**

Patient was diagnosed with Renal hyperoxaluria and patient was referred to the opthalmic department for opinion. Ophthalmic evaluation of the patient was done. Vision: 6/18, improving with pinhole to 6/6 in both eyes. Both eyes, anterior segment was normal. Fundus examination showed whitish yellow deposits of oxalate crystals superior and inferior to the disc. Palor of the Optic disc was present.

**Management:**

Diagnosis was confirmed, and patient is currently awaiting a combined kidney and liver transplantation from a cadaver donor, as isolated kidney transplantation is not advised.

**Discussion:**

Primary hyperoxaluria type I is an autosomal recessively inherited metabolic disorder caused by a deficiency of the peroxisomal enzyme alanine glyoxylate aminotransferase (AGT) in liver. The defective transamination of glyoxylate causes overproduction and urinary excretion of oxalate and glycolate causing recurrent calcium oxalate urolithiasis and nephrocalcinosis, end-stage renal failure, and systemic oxalosis with precipitation of calcium oxalate crystals throughout the body. In patients with the juvenile or adult variants the most common ocular abnormalities are crystalline spots or yellowish flecks in the posterior pole, and rare manifestations include macular black lesions, retinal perifoveal crystal deposition, retinal vascular occlusions, and neovascularisation. The diagnosis of Primary Hyperoxaluria should be suspected in a patient with recurrent kidney stones, normal urinary calcium and uric acid excretion, calcium oxalate crystals in the urine sediment, and marked hyperoxaluria in the absence of gastrointestinal disease or the ingestion of megadose vitamin C. The efficacy of treatment for Primary Hyperoxaluria depends upon early diagnosis and in minimizing renal calcium-oxalate deposition before advanced renal failure. Maintenance of high urine output, avoidance of high oxalate-containing foods, thiazide diuretics to diminish urinary calcium excretion have been tried. Intervention is required when stones obstruct the urinary tract. Percutaneous surgery or nephrostomy are preferred, since surgical removal may precipitate acute renal failure. Renal transplantation is life saving in oxalotic patients with end-stage renal disease, but it leaves the patient exposed to the risk of recurrent stone formation and nephrocalcinosis. Liver transplantation replaces the enzyme-deficient organ and may offer definitive treatment if combined with renal transplantation or performed before irreversible renal damage has occurred. A similar study was done by Small KW, Letson R, Scheinman J at the Department of Neurology, Duke University, Durham, NC in the year 1990.
The study included 24 patients with primary hyperoxaluria. Eight (30%) of the 24 patients with primary hyperoxaluria exhibited a bilaterally symmetrical oxalate retinopathy. The abnormalities were predominantly confined to the posterior pole and ranged from many small (100- to 200-microns) subretinal black ringlets to single large (2- to 3-disc diameter) geographic lesions. In 3 of the 8 patients with oxalate retinopathy, diffuse optic disc pallor was evident.

References.