

# My kidneys just stopped working: Adult onset end-stage renal disease from rare metabolic disorder

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## Objective

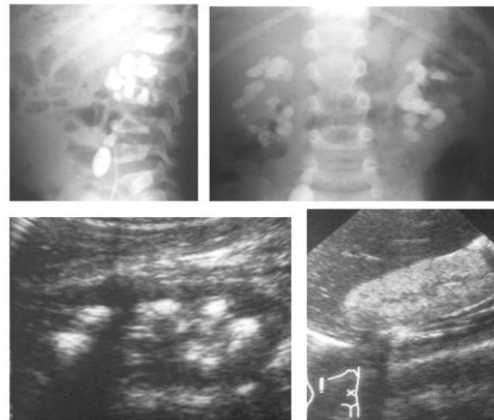
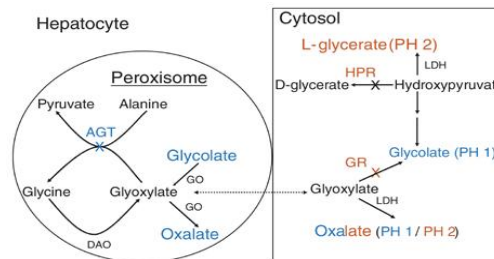
To introduce primary hyperoxaluria as a cause of end stage renal failure which presents with diffuse systemic manifestations

## Background:

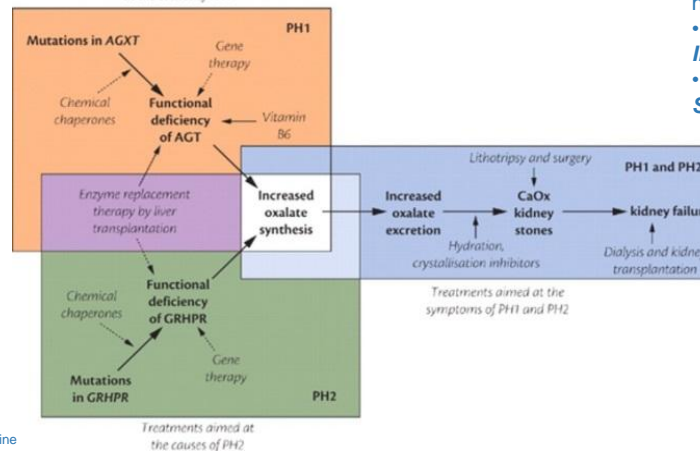
Primary hyperoxaluria (PH) is a rare metabolic disorder caused by defects in pathways of glyoxylate metabolism that causes increased oxalate production. PH type 1 (80%, most common) is due to defects in the gene that encodes hepatic peroxisomal enzyme alanine:glyoxylate aminotransferase (AGT), which converts glyoxylate to glycine. Early renal failure is a common outcome due to calcium oxalate nephrolithiasis formation and nephrocalcinosis. Untreated hyperoxaluria can result in systemic oxalosis causing damage to the heart (arrhythmias, cardiomyopathy), nerves (neuropathy), blood vessels (gangrene), kidneys (nephrolithiasis, nephrocalcinosis), eyes (retinal oxalate deposition), and joints (synovitis).

## Case Report:

48 yo white male presented through the ED with dyspnea, diffuse arthralgias (wrists, ankles, and knees), sick sinus syndrome s/p recent cardiac arrest and intra-cardiac device (ICD) placement, and end-stage renal disease for 4 months with a history of nephrolithiasis since childhood. He previously had received all care through an outside hospital where he was followed for rapidly progressive renal failure requiring dialysis after having minor renal insufficiency with serum creatinine of 2.0 only 8 months prior. Work up including physical and laboratory evaluation showed elevated serum oxalate level of 59 umol/L (normal < 27 umol/l) despite hemodialysis. After diagnosis of PH his symptoms improved with six times weekly hemodialysis. Initial evaluation for simultaneous liver-kidney transplant was initiated for definitive treatment.



Treatments aimed at the causes of PH1



## Discussion:

PH is a rare disease affecting 1 to 3 persons per 1,000,000. Further, cases of adult onset end-stage renal failure from this disease are exceedingly rare based off literature review. Despite having classic constellation of symptoms of this crystallopathy, his diagnosis was delayed. Clinical suspicion must be maintained for hereditary causes of renal failure in adults when usual work up remains negative.

## Diagnosis

Diagnosis typically made in childhood after recurrent calcium stones and oxalate crystals in urine sediment. Renal ultrasound shows urolithiasis nephrocalcinosis. Urine studies showing marked hyperoxaluria in absence of GI disease. Measurement of 24 hour urinary oxalate is preferred for diagnosis but can be falsely low with decreased Gfr. With renal dysfunction plasma oxalate/creatinine ratio or liver biopsy to measure AGT activity can be used.

## Treatment

Goal -- reduction of urinary calcium oxalate saturation and oxalate production

- **Initial treatment**  
**High dose pyridoxine** (10mg to 300mg daily) which helps convert glyoxylate to glycine rather than oxalate
- **With Renal Dysfunction**  
**Intensive dialysis** (Daily 5-hour sessions)
- **Permanent Treatment**  
**Simultaneous liver and kidney transplant**

## REFERENCES

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