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A Stone Cold Diagnosis: A Rare Claudinopathy Hiding Behind Common Nephrolithiasis Requiring Renal Transplant in a Child

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Case Study : A 12 year old girl, with recurrent abdominal pain was found to have bilateral multiple renal calculi at 5 years of age and underwent ureteroscopy, laser lithotripsy with DJ stenting at 8 years. Child however remained symptomatic, parents resorted to native medications and lost follow up. At 11 years, during an intercurrent illness, her renal function tests were found to be deranged [Serum urea: 78mg/dl, Creatinine: 7.7mg/dl]. Ultrasound revealed bilateral multiple renal calculi, small right kidney and gross hydroureteronephrosis of left kidney. The child was in stage 5 CKD with metabolic bone disease. A diagnosis of Hyperoxaluria was made elsewhere and started on intermittent hemodialysis. On treatment, she had multiple episodes of seizures and resistant hypocalcemia. On presentation to us, the unlikely finding of hypomagnesemia in a CKD child (CKD classically has hypermagnesemia) causing treatment resistant hypocalcemia to an extent of requiring magnesium supplements made us reconsider the established diagnosis of Primary Hyperoxaluria. Genetic testing was done which revealed the rare mutation in CLDN 16 gene causing Familial hypomagnesemia and hypercalciuria with nephrolithiasis(FHHNC) – a rare and under-reported entity. In view of recurrent urinary tract infections and mass effects of the left kidney, child underwent left nephrectomy in January 2026. She has successfully undergone renal transplantation in March 2026. The notable observation in this case was its presentation as renal stone disease, a presentation seen in multiple commonly encountered conditions like hyperparathyroidism, RTA, Idiopathic hypercalciuria and Hyperoxaluria, thus evading diagnosis. The striking feature of hypomagnesemia was key in diagnosing FHHNC, a rare autosomal recessive tubular disorder characterized by the triad of hypomagnesemia, hypercalciuria and nephrocalcinosis. It is caused by mutations of Claudin 16 gene encoding proteins responsible for about 70% of magnesium and significant fraction of calcium reabsorption. Progressive renal failure is inevitable.

FHHNC Fig 1.jpeg