Shaping Tomorrow's Nephrology: Insight-Driven Kidney Care
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From Nephrolithiasis to Systemic Oxalosis: the First Genetically Confirmed Case of Primary Hyperoxaluria Type 1 in Kazakhstan

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Case Study : Introduction Primary hyperoxaluria type1 (PH1) is a rare inherited disorder of glyoxylate metabolism characterized by excessive hepatic oxalate production,leading to recurrent nephrolithiasis and progressive kidney failure. To the best of our knowledge, this report describes the first genetically confirmed case of PH1 diagnosed in Kazakhstan. Case Report A girl born in 2009 presented at age 9 with macroscopic hematuria and right flank pain. Computed tomography performed in June 2019 demonstrated bilateral staghorn calculi measuring 5.5 cm in the right kidney and 4.5 cm in the left kidney with reduced renal parenchymal thickness. Endoscopic stone removal with retrograde pyelolithotripsy and ureteral stenting was performed. Despite surgical removal of the stones and relief of urinary obstruction, kidney function deteriorated rapidly. Serum creatinine increased from 96 µmol/L at presentation to 420µmol/L within 5 months and subsequently to 912µmol/L at initiation of dialysis. The rate of decline appeared disproportionate to obstructive uropathy. Early bilateral staghorn nephrolithiasis, accelerated CKD progression, and positive family history prompted genetic investigation. In March 2020 compound heterozygous pathogenic variants in the AGXT gene (c.508G>A p.Gly170Arg; c.568G>A p.Gly190Arg) confirmed PH1. Kidney function continued to decline despite surgical management, and renal replacement therapy was initiated in 2020, 9 months after the first manifestation of the disease. Peritoneal dialysis achieved recommended adequacy targets, but residual kidney function was rapidly lost with development of anuria. Laboratory findings included anemia (hemoglobin 95g/L), hypocalcemia 1.66mmol/L, hyperphosphatemia 2.39mmol/L, and progressive secondary hyperparathyroidism 621pg/mL. Cardiac evaluation demonstrated dilated cardiomyopathy with left ventricular ejection fraction decreasing to 26%, improving to 35-41% with therapy. Conclusion This first reported case highlighted that persistent kidney function decline after stone removal should raise suspicion for PH1. Early recognition is crucial because definitive treatment requires liver-kidney transplantation, and increased awareness may improve future detection of this rare disease.

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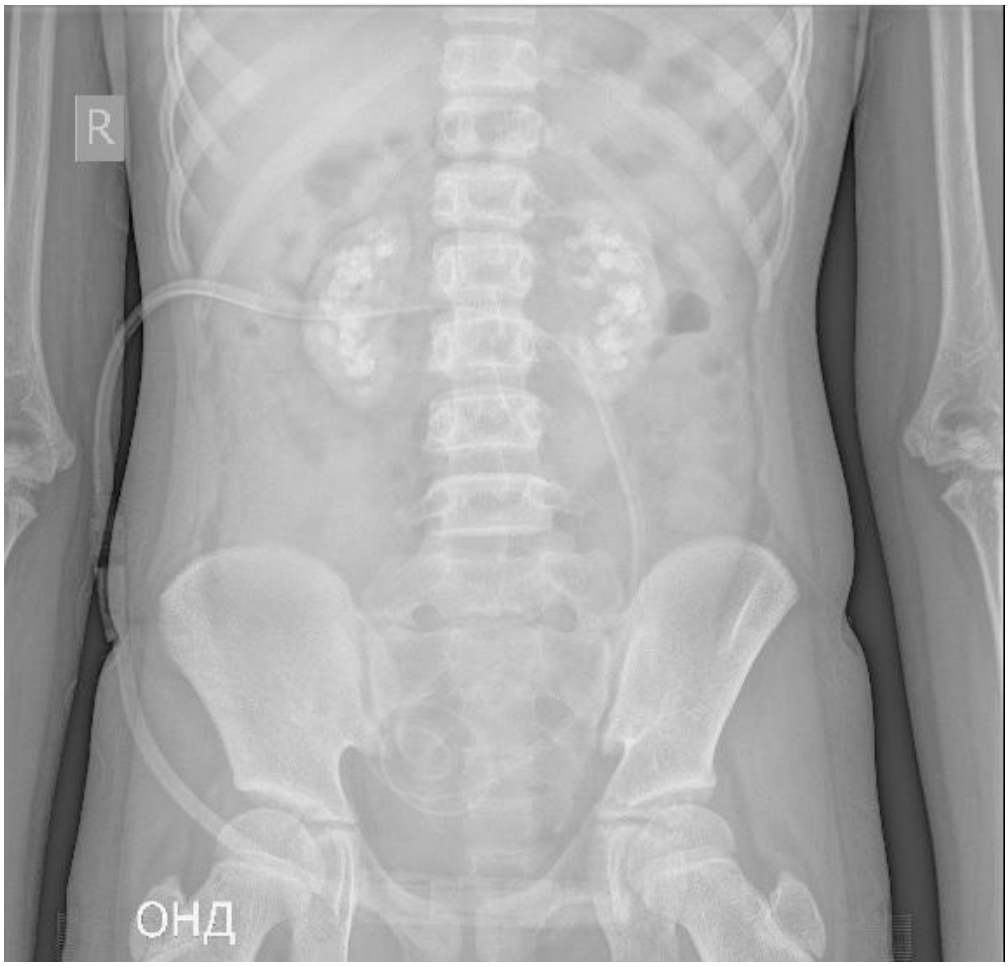
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