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Bilateral Medullary Nephrocalcinosis and Macular Dystrophy: Unmasking a Rare Tubulopathy

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Abstract

Early onset bilateral nephrocalcinosis often raises suspicion for inherited tubulopathies. Familial hypomagnesemia with hypercalciuria and nephrocalcinosis (FHHNC) represents one such entity. It is a rare autosomal recessive renal tubular disorder characterized by excessive urinary magnesium and calcium wasting, nephrocalcinosis, nephrolithiasis, and progressive renal dysfunction. Extra-renal features such as ocular abnormalities may also occur, particularly in type 2 disease. Herein, we present an 8-year-old girl who was referred to us for evaluation of bilateral nephrocalcinosis detected over the preceding 3 years. Clinical history revealed polyuria and polydipsia since early childhood. Ophthalmologic evaluation showed bilateral horizontal nystagmus, severe myopia, and bilateral macular dystrophy. Laboratory investigations demonstrated hypocalcemia, hypomagnesemia, hypercalciuria, hypermagnesuria, secondary hyperparathyroidism, and elevated serum creatinine. Imaging revealed bilateral medullary nephrocalcinosis in addition to bilateral renal and ureteric calculi. A provisional diagnosis of FHHNC type 2 was made. The patient was managed with oral magnesium supplementation, potassium citrate, and thiazide diuretics. This case highlights the importance of considering rare renal tubular disorders such as FHHNC in children presenting with bilateral nephrocalcinosis along with ocular abnormalities. Early recognition and supportive management are crucial to delay the progression of end stage renal disease which is the major cause of mortality in these patients. *[J Assoc Clin Endocrinol Diabetol Bangladesh, 2025;4(Suppl 1): S66]*

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