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Clinical presentations, genotypic spectrum, and outcomes of children with primary hyperoxaluria at Charlotte Maxeke Johannesburg Academic Hospital

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ABSTRACT

Background:

Primary hyperoxalurias (PHs) are a rare group of autosomal recessive disorders involving the overproduction of oxalate which results in renal calculus, progressive nephropathy, and eventual renal failure.

Objectives:

This study describes the demographics, clinical presentation, genotypic spectrum, determinants of disease severity and causes of death in patients diagnosed with PH, at a tertiary hospital.

Methods:

Retrospective descriptive review of patients with PH at a single unit, over 20 years.

Results:

Sixteen patients were identified with a median age at presentation of 7.1 years. Clinical presentations included nephrolithiasis and urinary tract infections (UTI) in six and end-stage kidney disease (ESKD) in five. Eight had a homozygous mutation c.335C>A (p.A112D) while four had a heterozygous mutation. The twelve mutations found were all on the AGXT gene for PH1. The median age of presentation of the four patients with heterozygous mutations was 5.7 years compared to 7.5 years for the homozygous mutation. Thirteen patients (81,3%) received renal replacement therapy (RRT). Eight patients were listed for transplant and six underwent a combined liver and kidney transplant (CLKT). Four patients were still alive at the time of this report and two patients had demised.

Conclusion:

Nephrocalcinosis was present in all patients, all of whom had PH Type 1. The genotypic spectrum correlated with initial presentations involving the urinary tract. The median estimated glomerular filtration rate (eGFR) was 4.2 ml/min/1.72m2 describing ESKD, however only five presented clinically in ESKD. This emphasizes the importance of screening, education, and earlier detection in our population.