Editorial

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Editorial on Primary Hyperoxaluria

Sophia Roberts

Managing Editor, Journal of Rare Disorders: Diagnosis & Therapy, United Kingdom

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***Corresponding author:** Sophia Roberts

raredisorders@medicineinsights.com

Tel: +44 7480022449

Managing Editor, Journal of Rare Disorders: Diagnosis & Therapy, United Kingdom.

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Editorial

Primary hyperoxaluria (PH) may be defined as a group of rare inherited disorders of the liver characterized by the overproduction of oxalate, an end-product of metabolism. High levels of oxalate are toxic because oxalate cannot be broken down by the human body and accumulates in the kidneys.

Primary hyperoxaluria are of 3 types of PH: type 1 (PH1), type 2 (PH2), and type 3 (PH3). In people with PH1, kidney stones typically begin to appear anytime from childhood to early adulthood, and ESRD can develop at any age. PH2 is similar to type 1, but ESRD develops later in life. In PH3, affected individuals often develop kidney stones in early childhood. PH1 affects 1 to 3 individuals per million, with a higher prevalence in some regions, such as the Middle East and North Africa [1,2].

Primary hyperoxaluria types 1, 2, and 3 are caused by the mutations in the *AGXT*, *GRHPR*, and *HOGA1* genes respectively. This lead to a decrease in production or activity of the respective proteins, which prevents the normal breakdown of glyoxylate. Mutations in *AGXT* and *GRHPR* gene result in an accumulation of glyoxylate, which is then converted to oxalate for removal from the body as a waste product [3,4].

Symptoms may include recurrent kidney stones; blood in the urine; and urinary tract infections. If left untreated, PH1 can result in end-stage renal disease, which is life-threatening.

When a person with PH1 has a kidney stone, symptoms can include: Flank pain, Painful urination, Urinary tract infections, Blood in the urine. Some individuals are not diagnosed until after their kidneys have failed and they require dialysis to help filter waste products from the blood. Patients can be diagnosed with PH1 at any age, but most individuals experience their first symptoms in early childhood and this can often results in end-stage renal disease, a life-threatening condition that prevents the kidneys from filtering fluids and waste from the body effectively. Consequently, the build-up of oxalate can lead to the deposition of oxalate crystals in the eyes, bones, skin, heart, and central nervous system, causing diminished vision, bone fractures, ulcers, heart failure, and other complications.

A diagnosis of PH is based upon identification of characteristic symptom. A variety of specialized tests includes Chemical analysis of urine samples, X-ray examinations, biopsy of affected kidney tissue, Examination of kidney stones and Molecular genetic testing.

The aim of treatment for primary hyperoxaluria type 1 (PH1) is to minimize calcium oxalate deposition and maintain renal function. General therapies and treatment for kidney stones may involve shock wave lithotripsy, percutaneous nephrolithotomy, and/or ureteroscopy. Dialysis may be used to treat individuals with PH.

For some individuals, it is consider to direct combined liver-kidney transplantation, sequential liver-kidney transplantation in case of severe systemic oxalosis or kidney transplantation in B6 responsive PH1 patients.

References

1 https://www.alnylam.com/patients/primary-hyperoxaluria/

2 https://rarediseases.info.nih.gov/diseases/2835/primary-hyperoxaluria-type-1

3 https://ghr.nlm.nih.gov/condition/primary-hyperoxaluria#

4 https://rarediseases.org/rare-diseases/primary-hyperoxaluria/