What is **Primary Hyperoxaluria**?

Primary hyperoxaluria (PH) is a family of ultra-rare, life-threatening genetic disorders that cause complications in the kidneys.

In patients with PH, the kidneys are unable to eliminate excess oxalate, which then accumulates in the kidneys and throughout the body.*

**Three known types of PH**

There are three known types of PH, each resulting from a mutation in one of three different genes. These genetic mutations cause enzyme deficiencies that manifest in the overproduction of a substrate called oxalate.

<table>
<thead>
<tr>
<th>Type</th>
<th>Genetic Mutation</th>
<th>Enzyme Deficiency</th>
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</thead>
<tbody>
<tr>
<td>PH1</td>
<td>AGXT</td>
<td>AGT</td>
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<tr>
<td>PH2</td>
<td>GRHPR</td>
<td>GR/HPR</td>
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<tr>
<td>PH3</td>
<td>HOGA1</td>
<td>HOGA</td>
</tr>
</tbody>
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**Signs and Symptoms**

Below is a list of potential signs and symptoms of PH:

- Kidney stones
- Kidney damage
- End-stage renal disease and injury to other organs
- Blood in the urine
- Urinary tract infections
- Retinal calcifications, resulting in vision problems
- Cardiac failure
- Arrhythmia
- Bone fractures
- Skin calcifications, resulting in painful skin nodules and necrosis

* The estimated genetic prevalence rates for PH imply more than 16,000 patients in the United States and European Union have the disease:

  - PH1: 1 in ~120,000
  - PH2: 1 in ~197,000
  - PH3: 1 in ~79,000
Current Treatment Options

Currently, there is no approved specific therapy for the treatment of PH. Patients are limited to using hyperhydration and medication to attempt to increase solubility of oxalate in urine.

Patients with severe PH may require regular dialysis and a dual liver-kidney transplant. Transplants are major surgical procedures, and subsequently patients must take immunosuppressant drugs for the rest of their lives.

References


Williams EL, Bockenhauer D, van’t Hoff WG, et al. The enzyme 4-hydroxy-2-oxoglutarate aldolase is deficient in primary hyperoxaluria type 3. Nephrology Dialysis Transplantation 2012; 27(8), 3191-3195.