

Diagnosing Hyperoxaluria

The signs of hyperoxaluria may not always be noticeable; they can go unrecognized or be confused with the signs of other diseases. Because of this, diagnosis is often a challenge among patients with hyperoxaluria. It often takes several years or longer after experiencing the first symptom and before the patient receives an accurate diagnosis. So, what are the signs and symptoms that individuals should be aware of?

Signs & Symptoms

Although kidney stones are the most common and often the first symptom of hyperoxaluria, not all individuals with hyperoxaluria will have kidney stones. If you experience the below signs and symptoms, you should further investigate to understand the cause, because it could be hyperoxaluria:



Urinary Tract Infections

Kidney Stones as a Child, Even Only 1

Blood in Your Urine

High Levels of Oxalate

Growth Failure

Failure to Thrive

Recurrent Kidney Stones

Nephrocalcinosis

Kidney Failure

Diagnostic Process

Your healthcare provider will conduct a thorough physical exam, including review of your medical history and discussion of your diet. The below tests are used to diagnose hyperoxaluria:



URINE TESTS: measures oxalate and other metabolite levels in the urine.



BLOOD TESTS: shows kidney function, as well as oxalate levels in the blood



STONE ANALYSIS: determines the composition of kidney stones that were either passed or surgically removed



KIDNEY X-RAY, ULTRASOUND, OR COMPUTERIZED TOMOGRAPHY (CT) SCAN: checks for any kidney stones or calcium oxalate deposits (where they should not be)



Importance of Genetic Testing

If you suspect you have hyperoxaluria, it may be possible that it could be Primary Hyperoxaluria (PH) or Enteric Hyperoxaluria (EH). To diagnose PH, genetic testing may be performed for screening. Since PH is an autosomal recessive disease that is passed down within families, it's important for all siblings in your family to be tested. EH is not genetic, so genetic testing for EH does not work. To diagnose EH, physicians examine patients' dietary intake and bowel habits. Further screening should be pursued to determine if you have other forms of hyperoxaluria, including EH or unknown types."

For information on no-cost genetic testing opportunities, please visit OHF at www.ohf.org or contact by email at info@OHF.org.

Treatment Options and Next Steps

Currently, there is one approved therapy for patients with PH1. Having treatment for those with PH1 is encouraging, but there is still much more work to do. OHF is forging ahead with great energy and fervor to find effective treatments and a cure for all patients living with all PH and EH. Read more about the approved therapy and ongoing clinical trials on the OHF website at www.ohf.org/clinical-trials.

If you suspect that you have any of the signs and symptoms associated with hyperoxaluria, you should discuss with your healthcare provider. Treatment depends on the type, symptoms, and severity of hyperoxaluria and how well you respond to treatment. For more information about diagnosing hyperoxaluria, visit www.ohf.org.