What is Hyperoxaluria?

Hyperoxaluria Awareness Week 2020

Hyperoxaluria occurs when your body produces too much oxalate in your urine. High levels of oxalate are toxic because oxalate cannot be broken down by the body and accumulates in the kidneys, causing kidney stones.

Primary hyperoxaluria can present at any age and at any time. Historically, PH has been underdiagnosed. Recurrent kidney stones in adults or any kidney stone in a child is usually the most common sign that you might have PH.

Hyper	oxal	uria
(too much)	(oxalate)	(in the urine)

What Causes Hyperoxaluria?

There are several causes of hyperoxaluria:

PRIMARY (COMES FROM WITHIN)–

which means that you have a genetic disorder

ENTERIC HYPEROXALURIA (ENTERING THE BODY)–

means that excess oxalate is absorbed into the gastrointestinal (GI) tract and then excreted in the urine. There are two forms of secondary hyperoxaluria:

- Fat malabsorption – several intestinal diseases, such as Crohn's disease or short bowel syndrome following bariatric and gastric bypass surgical procedures and/or complications, increase the absorption of oxalate in foods, leading to increased levels of oxalate
- Dietary – this means eating large amounts of foods high in oxalate can increase your risk of hyperoxaluria

IDIOPATHIC HYPEROXALURIA OR NO MUTATION DETECTED HYPEROXALURIA (UNKNOWN)–

which means that we are still unsure of what exactly causes this form of hyperoxaluria

There Are 3 Types of PH

In addition to enteric hyperoxaluria and idiopathic PH, there are 3 known types of primary hyperoxaluria (PH). PH is a family of rare, genetic liver disorders that can damage the kidneys. The liver normally makes proteins, called enzymes, that prevent the body from making too much oxalate. In PH, the liver doesn't create enough of this enzyme, or the enzyme doesn't work properly.

PH1	PH2	PH3
AGXT genetic mutation ↓ AGXT enzyme deficieny	GRHPR genetic mutation ↓ GR/HPR enzyme deficiency	HOGA1 genetic mutation ↓ HOGA1 enzyme deficiency

Signs and Symptoms

Although kidney stones are the most common and often the first symptom of PH, not all patients with PH will have kidney stones. Other signs and symptoms include:

- Urinary tract infection
- Kidney stones as a child, even only 1
- Growth failure
- Failure to thrive
- Recurrent kidney stones
- Nephrocalcinosis
- Kidney failure

Prevalence of Hyperoxaluria

It is estimated that about 200,000 to 250,000 people in the US have enteric hyperoxaluria. Approximately 1 to 3 of every million people have PH. There is even a higher prevalence of the disease in the middle east and in North African regions. It is estimated that half of the people with PH are undiagnosed. PH1 is the most common form of PH, accounting for approximately 80% of cases. PH2 and PH3 each account for about 10% of total PH cases.

Current and Future Treatment Options

Currently, there are no approved therapies for hyperoxaluria. Patients with hyperoxaluria are managed by very large volumes of daily fluid intake and surgery to remove recurrent kidney stones which does not address the underlying cause of hyperoxaluria. Patients with PH sometimes must undergo both liver and kidney transplants, which are major surgical procedures, and then they must then take immunosuppressant drugs for the rest of their lives to ensure they remain healthy.

Although there are a number of biopharmaceutical companies studying and developing therapies to potentially treat the hyperoxalurias. Read more about ongoing clinical trials on the OHF website at **www.ohf.org/clinical-trials**.

Oxalosis & Hyperoxaluria Foundation
STEPPING STONES TO A CURE

Accelerating research, providing hope empowering patients.

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