Hypophosphatasia and Your Kidneys—What to Expect

If you or a loved one has hypophosphatasia (*HPP*), you already know that it is a rare genetic disease that can affect the development of bones and teeth. In HPP, an inherited gene mutation interferes with the ability to produce alkaline phosphatase (*ALP*), an enzyme necessary for proper bone development. People with HPP might, for example, have frequent bone fractures, skeletal deformities, short stature, and/or premature loss of baby teeth.

What is less known is that people with HPP can be at a greater-than-typical risk for kidney damage— both because of the effect of insufficient ALP on the kidneys and as a result of high doses of certain pain medications. Although data is scant on the prevalence of kidney involvement, experts agree that it's essential for those with HPP to be aware of its signs, practice preventive measures, and get regular medical screenings to catch early indicators.

Why Does a Bone Disease Affect Kidneys?

ALP is required not only for normal bone development, but also for maintaining healthy kidney function. Ordinarily, calcium, phosphorus, and vitamin D are flushed out through the kidneys via a process that involves ALP. **Low ALP impedes the normal process**, causing these minerals and vitamins to accumulate and setting in motion a chain of physiological events that can result in kidney malfunction.

What's more, if you treat your HPP-related bone pain with nonsteroidal anti-inflammatory drugs (*NSAIDs*), as is common, you may inadvertently amplify the assault on your kidneys and hasten kidney damage.

HPP-Associated Kidney Problems

People with HPP can be predisposed to kidney stones. Because low levels of ALP hamper the ability of calcium to exit the kidneys, calcium accumulates and can form into kernels—stones—that get stuck in kidney ducts. Besides being very painful, chronic **kidney stones can lead to progressive kidney damage**—which, leads to **high blood pressure**, which, in turn, further damages the blood vessels of the kidneys. If you have high blood pressure from causes other than HPP (*family tendency, obesity, etc.*), it can exert an additive effect on the kidney-related impact of low ALP. If not properly and quickly addressed, **progressive kidney damage can lead to chronic kidney disease**.

What to Watch For

Several symptoms are early warnings of potential kidney involvement. If you experience any of these, contact the doctor managing your HPP. They include:

- **Burning or pain upon urination**
- **Blood in the urine**
- **Kidney pain** *(in the sides or middle to upper back on either side of the spine)*
- **Frequent urination**

Several signs of potential kidney involvement can be found through regular medical screening. They include:

- **High blood pressure**
- **High serum calcium** *(hypercalcemia)*
- **High urine calcium** *(hypercalciuria)*
- **Serum creatinine**
- **High serum vitamin D**
- **Ultrasound of the kidney**
- **High serum phosphorus** *(hyperphosphatemia)*

Based on early-warning signs or symptoms, the physician may refer you to a nephrologist (*kidney specialist*) for further evaluation and monitoring, which may include additional laboratory testing and ultrasound imaging.

How to Protect Your Kidney Health

- **Good water intake** to flush out high levels of phosphorus and calcium and minimize chance of crystal/stone deposits in the kidney
- **Limit NSAIDS to no more than three times per day**
  chronic use of NSAIDs can lead to kidney damage, increasing the damage from HPP
- **Normal, age-based calcium intake - avoiding high calcium intake** — too much calcium and vitamin D can cause renal complications — as the medical professionals move away from supplementing those with HPP with extra calcium and vitamin D, individuals with HPP should have fewer kidney problems.
- **Normal vitamin D intake**
- **Control your blood pressure**
- **Annual kidney assessment**, which should include blood pressure check, urinalysis, and serum creatinine
- **Regular DEXA imaging and bone-lab monitoring**, which should include parathyroid hormone and vitamin D status

Learn more about HPP and navigating life with HPP at SoftBones.org.
You can also contact Soft Bones at info@SoftBones.org

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Finding the Key to HPP
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