

# Kidney Stones are Common in Individuals with Pseudoxanthoma Elasticum

Douglas Ralph<sup>1</sup>, Rina Allawh<sup>1</sup>, Ian F. Terry<sup>2</sup>, Sharon F. Terry<sup>2</sup>, Jouni Uitto<sup>1,3</sup>, Qiaoli Li<sup>1,3</sup>

By Catherine Troutman and Georgia Boley

<sup>1</sup>Department of Dermatology and Cutaneous Biology, The Sidney Kimmel Medical College, and The PXE International Center of Excellence in Research and Clinical Care, Thomas Jefferson University, Philadelphia, PA 19107, USA

<sup>2</sup>PXE International, INC., Washington, DC 20008, USA

<sup>3</sup>Jefferson Institute of Molecular Medicine, Thomas Jefferson University, Philadelphia, PA 19107, USA

**P**seudoxanthoma elasticum (PXE) is a rare genetic disorder caused by mutations in the ABCC6 gene. People with PXE have signs of the disease in their skin, eyes, and vascular system. Sometimes people or their doctors suspect that other organs are affected. This is the case for the kidneys. People reported to PXE International and to their doctors that they had kidney stones. PXE International decided this might be associated with PXE and decided to conduct a study.

## What did we do?

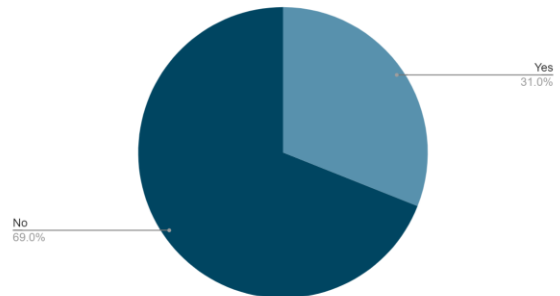
PXE International enlisted Douglas Ralph, Rina Allawh, Jouni Uitto, and Qiaoli Li from Thomas Jefferson University. We recruited 563 individuals to participate in the study. Participants were asked to complete a survey. The PXE International survey asked, "Have you ever had a kidney stone? How old were you when you first experienced a kidney stone? and How many times have you passed a kidney stone?". The researchers examined questionnaires and analyzed the data.

## What are the results?

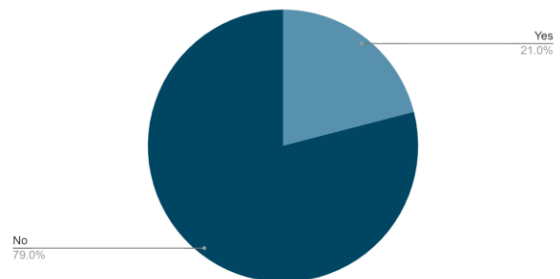
We found that 23.4% of people with PXE had kidney stones. Roughly 31% of the men that responded to the question stated they

experienced kidney stones, while 21% of women experienced kidney stones.

Percentage of Male PXEers who Experienced Kidney Stones



Percentage of Female PXEers who Experienced Kidney Stones



## How does this compare to the general population?

It is important to understand whether these kidney stones are caused by having pseudoxanthoma elasticum (PXE) or whether they are caused by something else. For example,

if we said 80% of people with PXE have brown hair and 20% have blond hair — is this caused by PXE? We would look at the general population (from the same region) and compare the PXEers to the general population and find that it was not being affected by PXE that caused hair color. Therefore, a good question is: are kidney stones found more often in PXEers than in the general public. And so, the researchers compared how common kidney stones are in the PXE population compared to the United States (US) general population.

A good reference for the average US population is something called The National Health and Nutrition Examination Survey (NHANES). It is a famous study that has gone on for many years in the US. These surveys are conducted by the Centers for Disease Control and Prevention (CDC) and investigate a large variety of health topics. NHANES had 28,629 participants 20 years or older in the data set the researchers analyzed. The researchers used kidney stone data from 2007 to 2016. The responses to the question, "Have you ever had a kidney stone?", were particularly interesting for this study. The individuals who were 20 years of age or older when they took the PXE International survey were compared to the NHANES population.

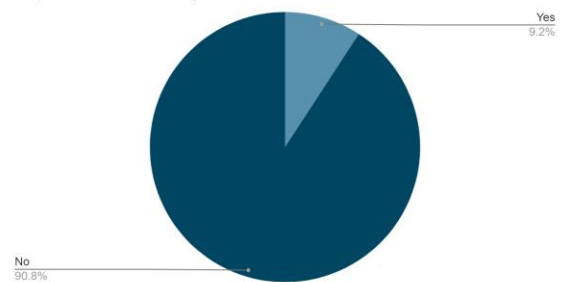
9.2% of the NHANES survey respondents had experienced kidney stones. In contrast, 23.4% of the PXE group reported experiencing kidney stones. PXEers are 3 times more likely to develop kidney stones.

In the PXE population, 17.8% of the PXE respondents experienced their first kidney stone before the age of 18. It is difficult to determine how common kidney stones are for individuals 18 and younger in the general population. However, it appears that PXEers get kidney

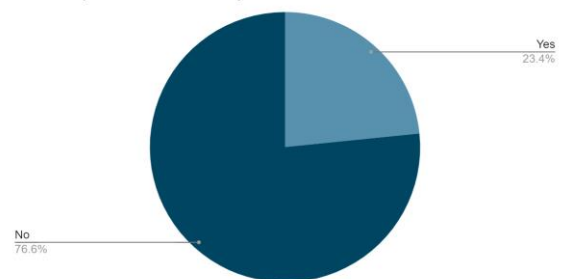
stones earlier and more often than people without PXE. Males are more likely to get kidney stones in the general population and so that is also true in the PXE population.

This study showed that people with PXE experience kidney stones more often than the general population. This does not mean that all people who have PXE will get kidney stones. In fact, about 2 out of 10 PXEers will. We do not know why people with PXE get more kidney stones, but we can guess that having low pyrophosphate allows more stones to build up.

Percentage of Respondents to the NHANES Survey Who Experienced a Kidney Stone



Percentage of Respondents to the PXE International Survey Who Experienced a Kidney Stone



### What else do we need to study?

Overall, individuals with PXE have an increased risk of developing kidney stones. The current Phenodex Score, which PXE International created to score your skin, eye, vascular, cardiac, and gastrointestinal signs/symptoms, does not

include looking at kidney functions. We think that it might be important to include this in the Phenodex.

**What can you do?**

- Make sure you hydrate enough, particularly when you exercise. This is probably the number one preventative.
- Calcium is Not the Enemy. In fact, too little calcium causes kidney stones! So make sure you have enough calcium in your diet.
- Eat less sodium. Too much salt in the urine prevents calcium from being reabsorbed from the urine to the blood. This causes high urine calcium, which may lead to kidney stones.
- Eat fewer oxalate-rich foods. Some kidney stones are made of oxalate, a natural compound found in food, that binds with calcium in the urine to form kidney stones. Limiting oxalate-rich foods may help prevent stones from forming. Foods high in oxalate are spinach, chocolate, sweet potatoes, coffee, beets, peanuts, rhubarb, soy products, and wheat bran.
- Eat less animal protein. Foods high in animal protein are acidic and may increase urine acid. High urine acid may cause both uric acid build-up and calcium oxalate kidney stones.

---

[\*More information about kidney stones\*](#)

[\*More information about pseudoxanthoma elasticum\*](#)

[\*\(PXE\)\*](#)

[\*Join the PXE Registry\*](#)