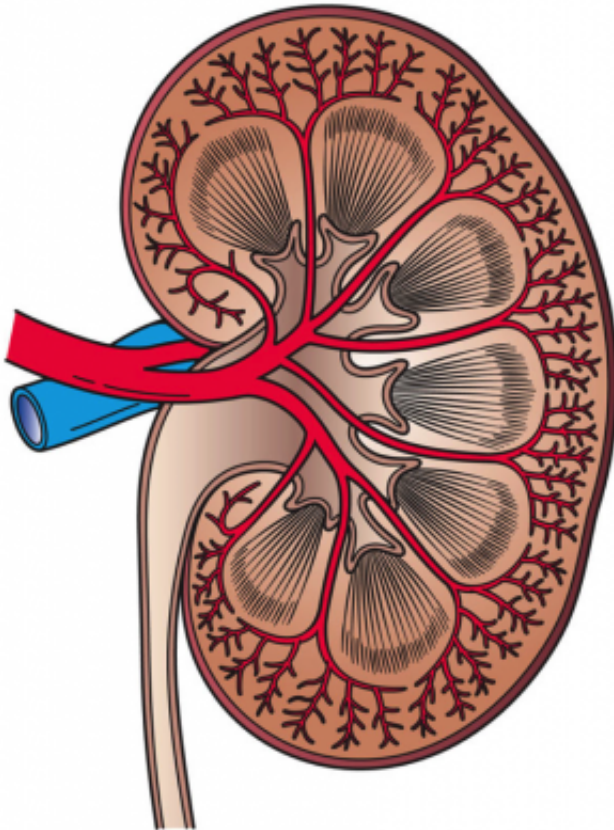


# Genetic link to kidney stones identified

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This image shows a cross section of a kidney. Credit: Holly Fischer/Wikipedia

A new breakthrough could help kidney stone sufferers get an exact diagnosis and specific treatment after genetic links to the condition were identified.

Academics from Newcastle and Harvard universities have found that

many cases of kidney stones have a [genetic cause](#) - meaning that specific early treatments can be given and other [family members](#) can be screened for the disease.

Kidney stones are a serious condition which affects one in ten people in the UK. Kidney stones are formed when minerals in the urine collect together to form blocks of [hard material](#). They are usually a few millimetres wide and impair the function of the kidney, lead to serious infections and blockages of the kidney.

They can cause [extreme pain](#) as well as long term damage to the kidneys. The new study, by Dr John Sayer and his team, has just been published in *Journal of The American Society of Nephrology*. The team studied the genes of 272 patients and found specific genetic mutations in 15% of cases, a number far higher than was previously thought.

These findings will enable doctors to manage and treat some patients with kidney stones more effectively.

Dr Sayer said: "Previously it was thought that about 1% of kidney stone cases were caused by genetic disease. But this study shows that genetic causes of stones are far higher and a significant proportion of cases are related to our DNA. That means we can identify patients and family members who are at risk of developing stones and hopefully intervene to prevent this very painful and serious condition.

"Nearly everyone who suffers from kidney stones will have further episodes of kidney stones. Current treatments include medical and surgical options. In light of this research we now aim to direct medical treatment at the precise cause of the condition to avoid recurrent stones and the multiple surgical procedures which are often needed."

## Kidney damage

Simon Moore, 38, of Workington, West Cumbria, has had kidneys stones for the past 20 years, caused by a condition called Cystinuria. His condition was so severe that he has had to have several operations which have caused further damage to his kidneys.

He said: "They started when I was 16 years old. I woke up in excruciating pain at 6am in the morning. I went to see the GP, he said it could be a [kidney stone](#), so I had an x ray which confirmed there was a blockage.

"The blockage was in my left kidney, it was about the size of my thumb and it meant I had to have hospital treatment for two months. Eventually I had to have quite major surgery. Both kidney's were affected.

"The condition is genetic in my case but no one else on my family ever had it. It's really hard to describe how horrible the pain is. You don't feel real.

"I have a lot of [kidney damage](#). I've had nearly 50 operations because the stones keep coming back. Drinking a lot of water does help but it just slows it down. Anything that could help prevent others in my family down the line having to suffer what I have would be fantastic.

"The charity International Cystinuria Foundation has been fantastic it has really supported me through difficult times."

Provided by Newcastle University

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