

# What is cystinuria?

**This leaflet explains about cystinuria, including its symptoms and treatments. If you have any further questions or concerns, please do not hesitate to contact the stone nurse.**

## What is cystinuria (cystine-in-the-urine) and why do I have it?

Cystine is one of the amino acids that make up proteins in the body. Most people's kidneys break down these amino acids and re-absorb them into the body. In people with cystinuria, the kidneys are unable to do this. Cystine builds up in the urinary tract, causing crystals to form and eventually turn into kidney stones.

Cystinuria is a genetic disease (this means you were born with it) but a stone can appear for the first time at any age, from when you are a baby right through to your sixties. Most people with cystinuria get stones in their twenties or thirties.

## How common is cystinuria?

One in 100 of adults and 6 to 8 in 100 of children with kidney stones have cystinuria. Around one in 2000 people in the UK are affected by cystinuria. It is more common in Caucasians (people of European origin). It affects men and women equally although men often have a more severe form of the disease.

## What are the signs and symptoms?

The most common symptom of stones is known as 'renal colic', which is severe back pain in the area over the kidneys. Some people have blood in the urine (haematuria) or urinary tract infections (UTIs). Some people may find out they have cystinuria when they are tested because a family member has it.

Approximately three quarters of people have stones in both kidneys. Although you may pass your stones when you pass urine or have treatment to get rid of them, they often come back. You will need to take steps to reduce the chance of getting kidney stones for the rest of your life. See the section 'Is there anything I can do to help myself?' on page 2 for more information.

## Do I need any tests to confirm the diagnosis?

When somebody gets a kidney stone or gets recurrent kidney stones, we will check for cystinuria. This involves testing for cystine in a urine specimen; this may involve collecting all of your urine over a 24 hour period or testing a fragment of stone.

## **Will my kidneys be affected?**

Kidneys have many important functions including filtering waste products from the blood, controlling blood pressure and producing some hormones. It is possible that having cystinuria could affect how efficiently your kidneys work. We have a kidney doctor (nephrologist) in the clinic to help monitor this and reduce the risk of serious kidney damage.

## **Is there anything I can do to help myself?**

You cannot change the fact that you have cystinuria but you can take some measures to reduce the chance of getting kidney stones.

### **Diet**

Drink plenty of fluid, so that you produce over 2.5 litres of urine each day. Ideally you should wake up once at night to pass urine. Drinking this amount of fluid can prevent stones in up to a third of people.

Cystine stones are formed in urine that has a high acid content so altering your diet can help to keep the urine less acidic. Cystine is formed from methionine (another amino acid) which is present in high quantities in eggs, meat and fish. Our dietitian can discuss your diet with you and help tailor a diet to your particular needs. Altering your diet to reduce your salt intake will also prevent stones forming. .

Levels of acidity in your urine are measured using a pH scale and the ideal pH you should be aiming for is 7.5. We will provide you with the pH sticks and explain how to use them. Then you can monitor your own pH at home.

### **Medicines**

Some people still get stones despite altering what they eat and drink. They may need to start taking tablets.

There is a medicine called potassium citrate that can help reduce the acid in your urine but this should only be prescribed after consulting your urologist.

Patients may be given penicillamine tablets which act by combining with the cystine to form a substance that dissolves in urine and prevents a stone forming. If you are already taking penicillamine then you should continue to, however some people experience unpleasant side effects and cannot take it. These patients are given Thiola® (alpha-mercaptopyrionylglycine or tiopronin) which has fewer side effects for some people than penicillamine. Tell your doctor if you are allergic to penicillin as that may mean that you will also be allergic to penicillamine.

## **What treatments are available?**

Cystine stones can be treated in exactly the same way as any other kidney stones and separate information leaflets are available on the different procedures below. Your doctor will discuss with you if these procedures are suitable for you:

**Extracorporeal shock wave lithotripsy (ESWL)** – this procedure aims to break your stones into smaller fragments that you then pass in urine. ESWL is successful for approximately half of patients with cystine stones. It is a day case procedure so you would usually not need to stay in hospital overnight.

**Ureteroscopy and laser treatment** – this is an operation to fragment and remove even large stones from inside. It requires an anaesthetic and usually involves a one night's stay in hospital, although it can be a day case. You may also have an internal "stent" placed temporarily which is a thin plastic drainage tube.

**Percutaneous nephrolithotomy (PCNL)** – this is a bigger operation that involves removing large kidney stones through a puncture hole made in your back. You should expect to stay in hospital for 2-3 days and you will need an anaesthetic.

**Open surgery** – this is very rarely done nowadays.

## Will my children get it?

Cystinuria is inherited (it runs in families). If you were to have a child with somebody who has cystinuria, then your child will also have cystinuria. If you were to have a child with somebody who carries a gene for cystinuria but does not have the disease, then your child has a one in two chance of having cystinuria. If you have a child with somebody who does not carry a cystinuria gene, then your children will not have cystinuria but they will carry the gene.

There are two main genes for cystinuria: SLC3A1 (type A) and SLC7A9 (type B). As explained above, you would usually need to have two of the cystinuria genes (one from each parent) to be affected with cystinuria. However, the SLC7A9 gene is more complicated as sometimes you can have the disease with only one copy of the gene (from one parent).

We offer genetic testing to our patients with cystinuria, so you will know the type of gene you have.

## Useful sources of information

w: [www.cystinuriaUK.co.uk](http://www.cystinuriaUK.co.uk)

w: [rarerenal.org/patient-information/cystinuria-patient-information/](http://rarerenal.org/patient-information/cystinuria-patient-information/)

### Contact us

If you have any questions or concerns about cystinuria, please contact the stone nurse via the bleep (Monday to Friday, 9am to 5pm). Call the hospital switchboard on 020 7188 7188 and ask for the bleep desk. Ask for bleep 0384 and wait for a response. This will connect you to the stone nurse practitioner directly.

Outside these hours in an emergency, please call the hospital switchboard on 020 7188 7188 and ask for the urology on-call doctor.

For more information leaflets on conditions, procedures, treatments and services offered at our hospitals, please visit [www.guysandstthomas.nhs.uk/leaflets](http://www.guysandstthomas.nhs.uk/leaflets)

### **Pharmacy Medicines Helpline**

If you have any questions or concerns about your medicines, please speak to the staff caring for you or call our helpline.

**t:** 020 7188 8748 9am to 5pm, Monday to Friday

### **Your comments and concerns**

For advice, support or to raise a concern, contact our Patient Advice and Liaison Service (PALS). To make a complaint, contact the complaints department.

**t:** 020 7188 8801 (PALS)      **e:** [pals@gstt.nhs.uk](mailto:pals@gstt.nhs.uk)

**t:** 020 7188 3514 (complaints)      **e:** [complaints2@gstt.nhs.uk](mailto:complaints2@gstt.nhs.uk)

### **Language Support Services**

If you need an interpreter or information about your care in a different language or format, please get in touch:

**t:** 020 7188 8815      **e:** [languagesupport@gstt.nhs.uk](mailto:languagesupport@gstt.nhs.uk)

### **NHS 111**

Offers medical help and advice from fully trained advisers supported by experienced nurses and paramedics. Available over the phone 24 hours a day.

**t:** 111

### **NHS Choices**

Provides online information and guidance on all aspects of health and healthcare, to help you make choices about your health.

**w:** [www.nhs.uk](http://www.nhs.uk)

### **Get involved and have your say: become a member of the Trust**

Members of Guy's and St Thomas' NHS Foundation Trust contribute to the organisation on a voluntary basis. We count on them for feedback, local knowledge and support. Membership is free and it is up to you how much you get involved. To find out more, and to become a member:

**t:** 0800 731 0319      **e:** [members@gstt.nhs.uk](mailto:members@gstt.nhs.uk)      **w:** [www.guysandstthomas.nhs.uk/membership](http://www.guysandstthomas.nhs.uk/membership)