NHS
Gloucestershire Hospitals
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Patient Information

Cystinuria and stone prevention

Introduction

This leaflet provides information for patients who have been diagnosed with the condition cystinuria and advice on how to try to reduce the risks of developing future stones.

What is cystinuria?

Cystinuria is a rare inherited condition of the kidney affecting about 1 in every 7,000 people. People with cystinuria have an increased risk of forming kidney stones. These stones are usually first noticed when a person is between 10 and 30 years old, although 1 in 4 people will have stone disease at an earlier age. Some people are not aware that they have cystinuria until after their first kidney stone.

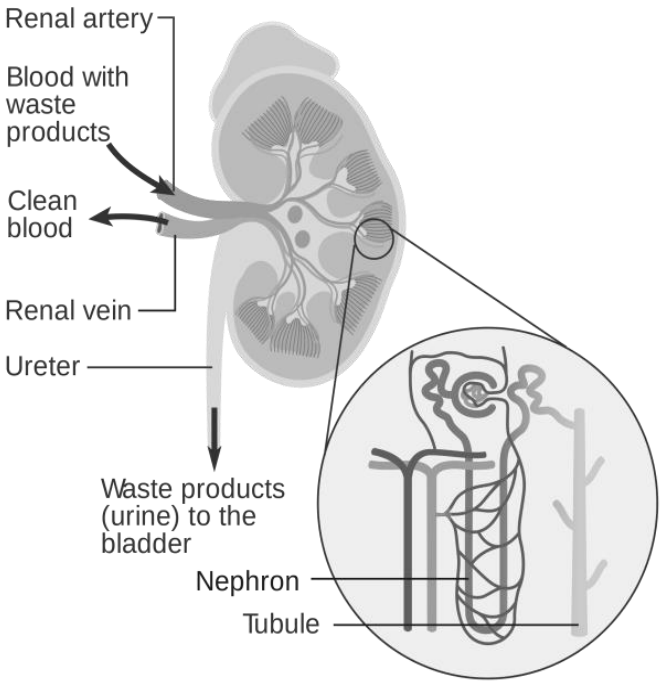

Figure 1: The normal function of a kidney

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Kidneys function like a sieve; they filter out waste for the body to get rid of. The kidneys also filter out things such as important nutrients and salts that the body will reabsorb.

Patients with cystinuria find it difficult to reabsorb certain amino acids (the 'building blocks' of protein) including one called cystine. When cystine is not absorbed back into the body, it stays in the kidneys and forms crystals. Most of these crystals are passed in the urine without you knowing. Sometime they can go on to form larger crystals which we call kidney stones.

What are the risks of cystinuria?

People who are diagnosed with cystinuria lead a normal life but are seen regularly in clinic to monitor the condition. People with severe cystinuria will be seen every 3 months. Evidence tells us that while treatment reduces the risks of forming kidney stones, it does not stop it completely; so the chances of having another kidney stone are high. People with cystinuria can develop 1 stone every 10 months.

Urinary tract stones, infections, and operations to remove stones are known to damage the kidneys. During your lifetime, there is about a 7 in 10 chance of your kidney's function getting worse because of cystinuria. There is a 5 in 100 chance that this will lead to needing some form of intervention. This risk can be reduced by making small changes to your diet and increasing the amount that you drink (see the 'Drinking more fluids' section).

As a result of the risk of damage to your kidneys, your kidney function will need to be monitored. This is done by having regular blood and urine tests. No other organs are affected by this condition.

What can you do to help?

There is no cure for cystinuria but with a combination of small lifestyle changes and sometimes the use of medications it is possible to lower the risk of your kidneys forming more stones.

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Drinking more fluids

Increasing the amount of fluids you drink will dilute (weaken) the cystine in your urine. This will reduce the chance of a stone forming. It is important, that when living with cystinuria you drink enough fluids to produce at least 5 litres (9 pints) of urine each day.

Useful measurements
1 cup (150mls)
1 mug (250mls)
1 tumbler (250mls)
1 pint (568ms)
1 ¾ pints (1000mls)

When increasing the fluids you drink try to avoid those high in sodium such as Lucozade® as these can encourage stones to form.

Citrus fruit juices such as orange and lemon juice are especially good as they can alkalinise (reduce the acidity of) your urine and lower your rate of further stones forming.

Night times are a period where you naturally tend to produce less, more concentrated urine. This also encourages stones to form so it is advisable that you drink in the evening, and take a glass of fluid to bed.

Tips
- Drink as much water as possible as part of your fluid intake.
- Carry drinks with you at all times.
- Drink with and between meals.
- Take a drink to bed with you.

Remember to drink more in hot weather or after exercise.

Having less salt

As well as benefits to the rest of your body, eating less salt lowers the amounts of cystine in your urine and will reduce the chances of further stones forming. Try to avoid adding extra salt to your food and read the labels on food products. You should try to keep your salt intake below 2 grams per day.

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Typical food and drink with high salt content:

Foods

- Processed meats such as ham
- Cheese
- Ready-made stocks/sauces
- Many condiments such as ketchup and soy sauce.

Drinks

- Energy drinks such as V® and Redbull®
- Sports drinks such as Lucozade®.

If in doubt, always check the nutritional information on food and drink packaging.

Other dietary changes

Cystine is formed by the breakdown of another amino acid (methionine). This is found in high quantities in animal protein; in both meat and dairy products. While a low methionine diet has not been proven to reduce the risks of stone formation, it is still advisable to eat no more than 2 small portions of meat or fish as well as 2 to 3 portions of dairy per day.

How else is cystinuria managed?

Urine alkalisation - Urine can be acidic or alkaline depending on what you eat and drink. It is known that alkaline urine helps to stop cystine forming kidney stones.

Medications like potassium citrate are given to people to make the urine more alkaline. If your urine is already alkaline, you do not need to take this as highly alkaline urine can encourage a different form of kidney stone to develop. As a result of this, your urine pH will be checked at your clinic appointments.

Chelation therapy - Sometimes a combination of lifestyle changes and careful control of urine pH are not enough to stop the formation of stones

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Medications called ‘chelation agents’ can be tried (such as penicillamine). These act by binding to cystine and lowering the chances of it forming stones. The choice of when, or if to start this medication, would need to be discussed with your urologist. This medication is associated with side effects including rashes, joint pains, blood disorders and changes to the function of your kidneys (called nephritic syndrome). If you begin to take this medication you will initially need regular blood tests to check your blood count and further urine tests. You will also need to start regular vitamins as levels of Vitamin B6 can be affected.

If you are pregnant, thinking of becoming pregnant or breast feeding, please tell your consultant as early as possible to discuss whether to continue taking these medications.

What to do if you develop stone symptoms

Pain from stones is referred to as renal colic. This is usually a pain that spreads from your back or side to the top of your thighs, called the groin. The pain often comes in waves and is usually a sign that a stone has passed into the ureter (the tube connecting a kidney to the bladder). Stones which are 5mm or less have a good chance of passing on their own.

If you have renal colic it is important to increase the amount you are drinking and take over-the-counter pain relief such as paracetamol. Renal colic can be very painful and you may need to be admitted to hospital.

You should seek medical advice if you have any signs of infection such as high temperatures, rigors (a sudden feeling of cold with shivering), unpleasant smelling urine and pain on passing urine or if the pain is not settling.

How can stones be managed?

There are many treatment options for stones. The treatment given will depend on the size of your stone and where it is.

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Available treatment

Shockwave Some stones can be broken up by using highly focused sound waves. This is the least invasive option but cystine stones are often hard and require several appointments. This option is limited to only certain stones.

Ureteroscopy This procedure requires you to be asleep so you will be given a general anaesthetic. A small camera is passed via the water pipe into the bladder and up the ureter to the stone where a laser is used to break it up. A temporary plastic tube (a stent) will be left in your ureter to allow healing after the procedure. Recovery is normally fast and patients are discharged the same or next day following the procedure. The stent is often left on a string and will be removed by a member of the Urology team a few days later.

Percutaneous nephrolithotomy (PCNL) This option is used for larger stones that cannot be removed in any other way. It requires a general anaesthetic. The procedure involves a small cut to the skin over the affected kidney to allow direct access through the kidney to the stone. After the operation, a plastic tube is left to allow the kidney to drain temporarily. You would need to stay in hospital for at least 2 to 3 days after this operation.

Is there a risk to your family?

Cystinuria is an inherited condition but it is important to understand that this does not mean that your children are bound to (or likely to) go on to develop the condition. If your partner has no family history of cystinuria, your child carries only a slightly higher risk than the general population with roughly a 1 in 84 chance of being affected.

Do

- Drink plenty of fluids (aim for 5 litres per day)
- Drink more when it is hot or you are exercising
- Check food and drink labels for salt content, you should have no more that 2g per day
- Eat a balanced diet.

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Do not

- Add salt to your food
- Eat large amounts of meat or dairy.

Further information

Cystinuria UK
Website: www.cystinuriauk.co.uk

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