CONDITION

Cystinuria

Cystinuria is an inherited condition that causes cystine kidney stones to form repeatedly.

WHAT IT IS
Inherited condition causing recurrent cystine kidney stones

BODY SYSTEM
Kidneys and urinary tract

MAIN SYMPTOMS
Side pain, blood in urine, nausea, urinary infections

WHAT CAUSES IT
Inherited changes affecting kidney amino acid transport

HOW IT IS DIAGNOSED
Urine cystine testing and laboratory stone analysis

MAIN TREATMENT
High fluid intake, dietary changes, medicines, stone removal

CAN IT BE CURED?
No - inherited transport changes remain

IS IT AN EMERGENCY?
Sometimes - fever with an obstructed urinary tract

LONG-TERM OUTLOOK
Recurrent stones, often manageable with lifelong care

Overview

Cystinuria is an inherited condition in which too much cystine passes into the urine. Cystine is made from two linked molecules of cysteine, an amino acid used to build proteins.

Normally, the kidneys filter cystine and return most of it to the blood. In cystinuria, this transport process does not work properly. Cystine builds up in the urine, where it can form hard crystals and kidney stones.

Stones can develop in the kidneys, the ureters that carry urine to the bladder, or the bladder. A stone may cause no symptoms until it moves, blocks urine flow, or contributes to an infection.

Cystinuria is lifelong. Treatment cannot remove the inherited change, but it can make cystine more soluble and reduce stone formation. Regular follow-up is important because stones often return and may affect kidney function.

Symptoms

Cystinuria usually causes symptoms only after a stone forms or moves. Symptoms can begin in childhood, adolescence, or adulthood.

Possible symptoms include:

Kidney stone pain often comes in waves and may change location as the stone moves. Pain severity does not reliably show the stone's size or whether it is dangerous.

Some stones are found during imaging for another reason. Children may have less specific symptoms, including abdominal pain, irritability, vomiting, or blood in the urine.

- Sharp or cramping pain in the side, back, lower abdomen, or groin
- Blood in the urine, which may look pink, red, or brown
- Nausea or vomiting during a painful episode
- Pain or burning when passing urine
- Needing to urinate more often
- Repeated urinary tract infections

Causes and risk factors

Cystinuria is caused by inherited changes in genes that help move certain amino acids through kidney cells. The genes most often involved are `SLC3A1` and `SLC7A9`.

The condition is usually inherited in an autosomal recessive pattern, generally meaning that a person inherits an altered gene copy from each parent. Inheritance can be more complex in some families, particularly with `SLC7A9` changes.

The transport problem also affects ornithine, lysine, and arginine, but these amino acids usually stay dissolved in urine. Cystine is less soluble, especially when urine is concentrated or acidic, so it forms stones.

Cystinuria is not caused by something a person ate or did. However, factors that can increase stone formation include:

Stone activity varies, even among relatives with the same inherited change. Some people have frequent stones, while others have long symptom-free periods.

- Not drinking enough to replace fluid losses
- Hot weather, heavy exercise, fever, vomiting, or diarrhoea

- High sodium intake, which can increase urinary cystine
- Urine that remains too acidic
- Missing treatment or follow-up

Diagnosis

Cystinuria may be suspected when stones begin young, recur, affect both kidneys, or occur in several family members. These features alone do not confirm it.

A passed or removed stone can be examined in a laboratory. Finding cystine confirms the stone type, so keeping a retrieved stone for analysis can be useful.

Urine may contain flat, six-sided cystine crystals. These strongly suggest cystinuria but are not present in every sample. Chemical screening or a measured urine cystine level can provide more evidence. A 24-hour urine collection may assess cystine, urine volume, acidity, sodium, and other stone risks.

Imaging shows a stone's location and size and whether it blocks urine flow. Ultrasound avoids radiation and is often used for monitoring. A CT scan provides more detail and may be needed during an acute episode or before a procedure.

Blood tests check kidney function. If infection is possible, urine is tested for bacteria. Genetic testing is not required for everyone but may help when the diagnosis is uncertain or detailed family counselling is wanted.

Treatment and management

Treatment aims to keep cystine dissolved, prevent new stones, slow existing stone growth, and remove stones that cause problems.

High fluid intake is central to treatment. The aim is to produce plenty of dilute urine throughout the day and night. The amount needed is individual, especially for children and people with heart or kidney problems. More fluid may be needed during heat, exercise, illness, pregnancy, or travel.

Reducing sodium can lower the amount of cystine entering urine. Some people are also advised to moderate animal protein, which can increase dietary acid load. Plans should still provide enough energy, protein, and nutrients, particularly for children.

Medicines that make urine less acidic may be prescribed because cystine dissolves better in alkaline urine. Potassium citrate is commonly used. Urine acidity can be checked at home with

test strips, while laboratory tests confirm whether treatment is working. Excessive alkalinisation can encourage other stone types, so monitoring is essential.

If these measures are insufficient, a cystine-binding medicine such as tiopronin or D-penicillamine may be considered. These medicines create substances that dissolve more easily, but they can cause significant side effects. Blood tests, urine tests, and regular review are required. Captopril is sometimes considered in selected circumstances but is not a standard substitute for more effective cystine-binding medicines.

A stone that is unlikely to pass, blocks urine, causes repeated infection, or produces persistent symptoms may need removal. Options include passing a small camera through the urinary tract to break up or retrieve the stone, or reaching a large kidney stone through a small opening in the back. Shock-wave treatment may be less effective because cystine stones are often hard.

Complications

Cystine stones can block a ureter and stop urine draining from the kidney. A blocked urinary tract becomes urgent if infection is also present.

Repeated stones and procedures may injure kidney tissue. Some people develop reduced kidney function or chronic kidney disease, although kidney failure is not inevitable.

Other possible complications include:

Treatment balances stone prevention against the risks of medicines and procedures.

- Recurrent urinary tract infections
- Kidney swelling caused by blocked urine flow
- Persistent pain or blood in the urine
- Scarring or narrowing within the urinary tract
- Side effects from long-term medicines
- Stress, sleep disruption, missed school, or time away from work

Prevention

Cystinuria cannot currently be prevented because it is inherited. Lifestyle changes cannot remove the underlying genetic change.

Many cystine stones can still be prevented or delayed by maintaining high urine volume,

limiting excess sodium, taking prescribed alkalinising or cystine-binding medicines consistently, and monitoring the urine.

Vomiting, diarrhoea, or fever can quickly reduce urine volume. Early attention to hydration may help, but persistent vomiting or inability to drink needs medical advice.

Relatives may discuss testing when there is a known family history. Genetic counselling can explain inheritance and testing options without assuming that a relative has the condition.

Living with it and what to expect

Cystinuria requires lifelong attention, even without symptoms, because the kidneys continue to release excess cystine into urine.

The course differs between people. Some have frequent painful episodes or repeated procedures, while others remain stable for years with fluids, dietary measures, and medicine. A quiet period does not mean the condition has gone away.

Follow-up commonly includes urine testing, kidney function tests, and imaging. These checks look for silent stones, growth, obstruction, treatment side effects, and kidney function changes. The schedule depends on age, previous stone activity, treatment, and imaging findings.

Daily management may involve carrying water, planning toilet access, checking urine acidity, and increasing fluid intake during heat or exercise. Overnight drinking may be discussed because urine becomes concentrated during sleep.

Most people can work, study, exercise, travel, and have families. Pregnancy can change fluid needs and affect which medicines are suitable, so treatment should be reviewed before or early in pregnancy.

Long-term care focuses on reducing stone formation and protecting kidney function, not only treating painful episodes.

When to get medical help

Seek emergency medical care for signs that a stone may be blocking an infected urinary tract:

An infected, obstructed kidney may require urgent drainage as well as antibiotics like amoxicillin or doxycycline .

Seek prompt medical advice for pain that cannot be controlled, repeated vomiting, difficulty keeping fluids down, visible blood in the urine, or new urinary symptoms. Children, pregnant

people, and people with reduced kidney function may need earlier assessment.

Attend routine follow-up to review urine results, imaging, treatment side effects, and difficulty meeting fluid or dietary goals. If you take a cystine-binding medicine, promptly report new swelling, rash, mouth ulcers, unusual bruising, or marked tiredness.

- Fever or chills with pain in the side or back
- Feeling severely unwell, confused, faint, or unusually drowsy
- Very little or no urine, especially with worsening pain
- Stone symptoms in someone with one functioning kidney

Common questions

Is cystinuria the same as ordinary kidney stones?

No. Kidney stones can contain different substances. Cystinuria specifically causes cystine stones because of an inherited kidney transport problem. Identifying the stone type matters because prevention and medicines differ from those used for common calcium stones.

Can cystinuria go away?

The inherited condition does not go away. Treatment can greatly reduce stone activity, and some people have long periods without a stone. Monitoring remains necessary because cystine continues to enter urine.

What should I drink if I have cystinuria?

Water is usually the main drink used to increase urine volume. The aim is plenty of dilute urine throughout the day rather than drinking a large amount at once. Individual needs depend on age, climate, activity, pregnancy, and heart or kidney health.

Can I pass a cystine stone naturally?

Some small stones pass without a procedure, but size is not the only consideration. Location, pain, infection, kidney function, and the degree of blockage also matter. A stone causing infection or significant obstruction should not be watched at home.

Will my children inherit cystinuria?

This depends on the gene involved and whether the other biological parent carries a relevant change. Children may inherit the condition, carry one altered copy, or inherit neither change. Genetic counselling can provide family-specific information.

Does cystinuria cause kidney failure?

Kidney failure is not inevitable. However, repeated obstruction, infections, stones, and

procedures can reduce kidney function. Long-term prevention and monitoring help protect the kidneys.

Sources

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