

Cystinuria

Information for women who have cystinuria during pregnancy or breastfeeding

The information provided below is for readers based in the United States of America. Readers outside of the United States of America should seek the information from local sources.

What is cystinuria during pregnancy?

Cystinuria is a genetic disorder in which there is excessive excretion through the kidneys of *cysteine*, which is an amino acid, a building block of proteins. In some forms of cystinuria, there also is excessive excretion of other amino acids in addition to cysteine. The cysteine accumulates in the urinary system and crystalizes into *cystine* stones (calculi) in the kidneys, ureters, and bladder (cystine is the result of two cysteine molecules linking together and becoming oxidized). Since it is a disorder resulting from mutations in two genes, if you are born with cystinuria, then you still have it when you are pregnant, so it can coexist with pregnancy.

How common is cystinuria during pregnancy?

Cystinuria is present in approximately 1 out of 10,000 people. This prevalence translates to pregnant women since the condition is passed down with what geneticists call autosomal inheritance, which means that it occurs roughly equally in males and females.

How is cystinuria during pregnancy diagnosed?

Typically, diagnosis is made after you experience an episode of urinary stones in which you pass a stone, or in which a stone is found in urine that is collected over time. In either case, a diagnosis is made when the stone is found to be a cystine stone. Diagnosis also can involve genetic testing for mutations of the genes *SLC31A* and *SLC7A9*. Additionally, your kidneys, ureters, and bladder may be imaged with ultrasonography, or with forms of magnetic resonance imaging (MRI) that image the urinary system. In some cases, computerized tomography (CT) scanning may be needed for a closer look, even though this exposes you and the fetus to some amount of ionizing radiation.

Does cystinuria cause problems during pregnancy?

Cystinuria causes the formation of stones, which can get stuck, causing obstruction of the urinary tract. This results in pain in the flank, upper abdomen, or the back. Typically, the pain advances to the lower abdomen or the groin, and you also may suffer from urinary urgency/frequency, nausea and vomiting, and blood in the urine. Often, [kidney stones](#) will eventually pass through into the urine, and the situation will resolve on its own. Otherwise, you will need treatment to break up the stone. If a stone does not pass and the condition is not treated, there is a possibility of severe complications, including an upper [urinary tract infection](#) that can damage the kidneys, an abscess (a concentrated big infection), sepsis (infection throughout the body), and various types of rupture and leaking of parts of the urinary system. Serious complications that arise from kidney stones may put you at increased risk for certain pregnancy complications, such as [preeclampsia](#) and gestational [diabetes](#) mellitus, and may increase the chances that you'll need a cesarean delivery.

Does cystinuria cause problems for the baby?

Some studies have suggested associations between urinary stones (most of which are not cystine stones) and pregnancy complications that can harm the baby, such as preeclampsia, [gestational diabetes](#), [premature rupture of membranes](#) (water breaks too early), low birth weight, and [spontaneous abortion](#) (miscarriage). Still, the findings are not consistent between different studies. On the other hand, if you develop a serious complication, such as sepsis, then the baby is in danger because your life is in danger.

If you have cystinuria and the baby's father does not have cystinuria and is not a carrier for it, then the baby will not have the condition but will be a carrier. If both you and the father have cystinuria, then the child will have cystinuria. If you have cystinuria and the father is a carrier but doesn't have the condition, then the baby has a 50 percent chance of having the condition and a 50 percent chance of being a carrier without having the condition.

What to consider about taking medications when you are pregnant or breastfeeding:

- The risks to yourself and your baby if you do not treat the cystinuria
- The risks and benefits of each medication you use when you are pregnant
- The risks and benefits of each medication you use when you are breastfeeding

What should I know about using medication to treat cystinuria during pregnancy?

Cystinuria can be treated with medications that make your urine more alkaline (less acidic), such as acetazolamide and potassium citrate. With this strategy, you must also limit your dietary intake of salt.

Although there has been some concern that acetazolamide might present a risk for the fetus, studies have not found much evidence supporting this concern, while potassium citrate is thought to be fairly pregnancy-safe. Outside of pregnancy, cystinuria also can be treated with a drug called D-penicillamine, which causes cysteine to swap for a type of cystine that dissolves more easily in the urine than the usual kind of cystine, but this drug is thought to pose a risk of birth defects.

Who should NOT stop taking medication for cystinuria during pregnancy?

If you are on medication to help prevent the formation of cystine stones by alkalinizing your urine, and if it has been working well for you, it is wise to continue the treatment. If you are on D-penicillamine, you should speak with your doctor about switching your treatment.

What should I know about choosing a medication for my cystinuria during pregnancy?

It is important to stay in communication with your health care provider as the release of new studies over time can change the outlook on the role of specific medications during pregnancy.

You may find Pregistrys expert reports about the medications to treat this condition [here](#). Additional information can also be found in the sources listed below.

What should I know about taking a medication for my cystinuria when I am breastfeeding?

Its generally considered fairly safe to nurse when taking acetazolamide, but it is recommended that you avoid breastfeeding if you are taking D-penicillamine.

What alternative therapies besides medications can I use to treat my cystinuria during pregnancy?

It is important for you to drink plenty of fluids. If you do form a cystine stone and it does not pass, there are procedures available, such as breakup and removal of a stone through an instrument called a ureteroscope into the bladder and kidney to remove or break up a stone. The standard procedure for the treatment of stones in non-pregnant people, extracorporeal shockwave lithotripsy (ESWL), is contraindicated in pregnancy. However, another procedure, called laser lithotripsy, appears to be safe and effective in pregnancy, particularly for ureteral stones.

What can I do for myself and my baby when I have cystinuria during pregnancy?

Follow the instructions of your physician and report any medication side effects to your physician. Consult with a genetic counselor and learn the inheritance pattern of the condition in order to be aware of your childs chances of inheriting the condition, or carrier status.

Resources for cystinuria in pregnancy:

For more information about **cystinuria** during and after pregnancy, contact <http://www.womenshealth.gov/> (800-994-9662 [TDD: 888-220-5446]) or contact the following organizations:

- [Healthline. Cystinuria.](#)
- [Urology Care Foundation. Pregnancy and Kidney Stones????????](#)

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General information

It is very common for women to worry about having a miscarriage or giving birth to a child with a birth defect while they are pregnant. Many decisions that women make about their health during pregnancy are made with these concerns in mind.

For many women these concerns are very real. As many as 1 in 5 pregnancies end in a miscarriage, and 1 in 33 babies are born with a birth defect. These rates are considered the background population risk, which means they do not take into consideration anything about the health of the mom, the medications she is taking, or the family history of the mom or the baby's dad. A number of different things can increase these risks, including taking certain medications during pregnancy.

It is known that most medications, including over-the-counter medications, taken during pregnancy do get passed on to the baby. Fortunately, most medicines are not harmful to the baby and can be safely taken during pregnancy. But there are some that are known to be harmful to a baby's normal development and growth, especially when they are taken during certain times of the pregnancy. Because of this, it is important to talk with your doctor or midwife about any medications you are taking, ideally before you even try to get pregnant.

If a doctor other than the one caring for your pregnancy recommends that you start a new medicine while you are pregnant, it is important that you let them know you are pregnant.

If you do need to take a new medication while pregnant, it is important to discuss the possible risks the medicine may pose on your pregnancy with your doctor or midwife. They can help you understand the benefits and the risks of taking the medicine.

Ultimately, the decision to start, stop, or change medications during pregnancy is up to you to make, along with input from your doctor or midwife. If you do take medications during pregnancy, be sure to keep track of all the medications you are taking.

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