



# Common investigations

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Cystinosis affects many different organs in your body. Therefore it may be necessary to attend further appointments to monitor and protect your other organs. This section provides a brief description of common investigations.

### Investigation: Blood tests

**Why do I need them?** Blood tests can be requested to assess a wide range of conditions and the function of organs. Common blood tests that you may have include:

- **Urea and Creatinine:** These are both waste products made by the body carrying out its normal day to day functions. Urea and Creatinine should be passed out in the urine and high levels in your blood may indicate problems with the kidney.
- **Electrolytes:** Electrolytes are the minerals that keep the body's fluid levels in balance and play an important role in helping the muscles, heart and other organs working properly. This blood test typically measures the level of sodium, potassium, chloride and bicarbonate in your blood.
- **Liver Function Tests:** This blood test measures the level of certain chemicals produced by your liver. The test can help assess if the liver is damaged and can help diagnose diseases associated with the liver.
- **Bone Profile:** This blood tests measures proteins, minerals, and enzymes involved in bone development. The results can help diagnose possible problems with the bone.
- **HbA1c:** This test measures your recent average blood glucose level. High levels may indicate diabetes.
- **Thyroid Function Test:** This blood test measures the level of the hormones produced by the thyroid gland. The results can help diagnose whether or not the thyroid gland is working properly.

**How often will I need blood tests?** This will depend on the results of the blood tests and you should be advised on how often to have

them checked by your doctor.

## Investigation: White cell cystine (WCC)

**Why do I need it?** Taking white cell cystine levels allows us to measure how much cystine is in your cells. The amount of cysteamine you are required to take depends on your WCC levels

**What is it?** To get a WCC level we need to perform a simple blood test. The blood must be taken 5-6 hours after your last dose of cysteamine.

**How often do I need it?** It is important to have your WCC levels taken regularly so that you are taking the appropriate dose of cysteamine. You should have your WCC levels checked approximately every 3 months.

**When can I expect the results?** The results can take a little while to come back because it is a specialist test. If you would like to be contacted to discuss your result please inform your healthcare professional.

## Investigation: Ultrasound scan (USS)

**Why do I need it?** Having an ultrasound scan can help detect abnormalities with your organs such as Liver, Kidney, pancreas, gall bladder etc.

**What is it?** An ultrasound scan is painless and safe. You'll lie down and a probe will be placed on your skin. Lubricating jelly will be used to ensure good contact between the probe and your skin. The probe sends out pulses of ultrasound to detect your organs. The scan will take between 15-30 minutes.

**How often do I need it?** The need for an ultrasound scan will be decided in clinic between you and your consultant.

**When can I expect the results?** Results are available quite quickly. Your doctor may write to you with the results or may discuss them with you in your next clinic appointment.

## Investigation: Echocardiogram

**Why do I need it?** Having an echocardiogram can give accurate information about the heart and can be useful in diagnosing any problems with the heart.

**What is it?** An echocardiogram is performed the same way as an ultrasound. Lubricating jelly is placed on your chest and a probe is used to send/receive ultrasound which will build a picture of your heart. It is painless and safe.

**How often do I need it?** The need for an echocardiogram will be decided between you and your consultant.

**When can I expect the results?** Your doctor may write to you with the results or they can be discussed at your next clinic appointment.

## Investigation: Lung function test

**Why do I need it?** Patients with cystinosis are at an increased risk of developing problems with their breathing as lung capacity becomes restricted due to prolonged high levels of cystine. Therefore it is important to monitor your lung function.

**What is it?** The specific test you have is called a spirometry and will require you to breathe in until your lungs are completely filled with air before you blow out as fast as you can into a measuring device.

**How often do I need it?** If you notice any changes in your breathing or you are beginning to feel increasingly short of breath you should inform your consultant. The need and frequency of lung function tests will be discussed between you and your consultant.

**When can I expect the results?** Results are available quite quickly. Your doctor may write to you with the results or may discuss them with you in your next clinic appointment.

## Investigation: Speech and Language therapist

**Why do I need it?** Patients with cystinosis can develop problems with the muscles around the tongue, lip and jaw area which may lead to difficulties in chewing/swallowing. Therefore it is important to see a speech and language therapist to help with any difficulties you may be having.

**What is it?** The speech and language therapist will spend some time getting to know you and any particular needs you may have. The therapist may offer you information, advice and exercises to help you.

**How often do I need it?** The need to see a speech and language therapist will be decided between you and your consultant. If you notice any new difficulties with chewing/swallowing it is important that you inform your doctor.

**When can I expect the results?** Any problems you have can be discussed with your therapist at your appointment.

### \*\*\*Important\*\*\*

If you have any concerns about the investigations you should discuss this with your healthcare professional. If you can't attend your booked appointment you should inform the applicable department and rebook as soon as possible. Missing appointments may be detrimental to your health.