What is primary hyperoxaluria

Primary hyperoxaluria is a rare condition characterised by the overproduction of a (PH) substance called oxalate.

Oxalate is a natural end product of metabolism and is not needed for any other bodily function. Approximately 90% of oxalate is removed by your kidneys.

Oxalate occurs naturally in certain foods and is also produced inside the body from a chemical called glyoxalate. Oxalate in the body is regulated by proteins called enzymes. In primary hyperoxaluria this enzyme is faulty, too much oxalate is produced and the body cannot get rid of it all. Oxalate doesn’t dissolve well and so tends to form solid crystals, especially when it combines with calcium. This causes kidney stones. This can damage the kidneys and stop further oxalate being excreted. Over time Oxalosis can occur, where the oxalate crystals that are not excreted by the kidneys are deposited in other organs such as the eyes, bones, skin, muscles, and heart.

Types of primary hyperoxaluria

Type 1, type 2, and type 3 primary hyperoxaluria are rare genetic conditions. Each type of primary hyperoxaluria is caused by a deficiency in a certain liver enzyme which causes large amounts of oxalate being produced.
How is it diagnosed?

Spot urine tests
A spot urine sample can be performed at any time of day and only requires a small amount of urine. This type of test can be useful for initial screening. However further tests are usually required to confirm diagnosis.

24 Hour urine tests
Diagnosis can be made by requesting specialist urine tests to see what is in your urine. You may be required to do a 24 hour collection which requires you to collect all the urine you pass over 24 hours into a container. To obtain an accurately timed specimen you should begin and end the collection period with an empty bladder. For example:

**Day 1, 08:00am** – Void and discard first urine of the day. Collect all following urine for next 24 hours.

**Day 2, 08:00am** – Void and collect first urine of the day and add to previously collected urine. Stop collecting urine from this point.

Stone analysis
Kidney stone analysis is a test done on a kidney stone that has been passed in the urine or removed surgically. Kidney stones can be made up of different chemicals depending on the condition.

Genetic testing
A genetic blood test can look for changes in your DNA that indicate a diagnosis of primary hyperoxaluria.
What are the symptoms?

Patients are usually unaware they have primary hyperoxaluria until they develop their first kidney stone. Renal colic (severe pain in the back, side and groin area) is often the first sign.

However not all stones cause renal colic. Some stones are small enough to pass without causing any pain.

What effect will the condition have on me?

You should be able to work and take part in daily activities as normal. You may need to take time off work or college to attend clinic appointments, and should be allowed unrestricted access to toileting facilities. If you are being denied either of these please let your doctor or nurse know.

There are complications with primary hyperoxaluria that all patients should be aware of, these are as follows:

Renal colic

Renal colic is when you experience moderate to severe pain caused by a stone travelling down the ureter (the tube that joins your kidneys to your bladder). The pain is noticeable in the lower back, the side, and moving into the groin.

Kidney/urine infections

Patients with primary hyperoxaluria are at an increased risk of developing kidney infections. The symptoms include pain when passing urine, and increased frequency or urge to pass urine.

Hydronephrosis

This is when the ureter becomes blocked and urine starts to backfill into the kidney. The kidney becomes enlarged and unable to function properly. The stagnant urine within the kidney can also cause a very serious infection. It is important that this is treated as soon as possible to prevent further complications.
Kidney damage

Over time large amounts of oxalate can cause irreversible damage to the kidneys. The severity of kidney damage depends on the type of primary hyperoxaluria you have and can be different for each patient. Additionally, repeated urine infections, kidney stones, blockages, and scarring from surgery can all have an effect on your kidney function. If your kidney function reaches a level at which they are no longer able to filter out waste products it may be necessary to start dialysis or have a kidney transplant. Your kidney function will be very closely monitored by your medical team.

Liver/kidney transplant

Some people with primary hyperoxaluria (PH) can develop progressive chronic kidney disease and some may require kidney dialysis or transplantation. In PH1 kidney a transplant is not a permanent cure as the oxalate will still damage the transplanted kidney. In some cases the only cure is to have a combined liver and kidney transplant. The transplanted liver will replace the missing enzyme which will return oxalate levels back to normal. The transplanted kidney will then be able to filter out waste products as normal. Having a transplant is a major procedure and should be discussed in detail with your medical team.
Who will look after me?
You will need to be managed by a specialist team of clinicians that may include the following:

- **Nephrologist**: A doctor who specialises in the kidneys who will monitor your kidney function and guide management to prevent new kidney stones forming. A nephrologist will also treat kidney failure and advise if your kidney function worsens. They will guide treatment if kidney dialysis or transplants might need to be considered.

- **Urologist**: A doctor/surgeon who specialises in the urinary tract (the kidneys, ureters, bladder and urethra) who will intervene and treat any oxalate stones that have formed.

- **Hepatologist**: A doctor who specialises in the liver who guides management if a liver transplant is necessary.

- **Nurse Specialists**: Senior specialised nurses will help coordinate your care and monitor your progress if your condition is stable.

What treatments are available?
Primary hyperoxaluria is a lifelong condition as there is currently no curative treatment available. Therefore treatment is aimed at preventing kidney stones and protecting kidney function. Treatment methods are listed below;

**High fluid intake**
Patients with primary hyperoxaluria are encouraged to drink high amounts of fluid. This will ensure that your urine is diluted which will decrease the concentration of oxalate in the urine. A lower concentration of oxalate reduces the chance of forming a kidney stone. The urine becomes more concentrated at night time and therefore we particularly encourage drinking extra before going to bed. The aim is to pass approximately 3 litres of urine a day, which may require drinking between 3.5–4 litres of fluid a day.

**Alkaline urine**
Urine can be acidic or alkaline depending on what you drink and eat. Patients with primary hyperoxaluria should aim to have alkaline urine.
This is because kidney stones are less likely to form in mildly alkaline urine. The common medications used to make urine more alkaline are:

- **Potassium Citrate**
  - Liquid solution that needs to be diluted
- **Effercitrate**
  - Fizzy tablet which needs to be dissolved in water

**Magnesium supplements**

Magnesium can help by binding with the oxalate, therefore preventing the oxalate from binding to calcium and forming stones.

**Vitamin B (pyridoxine)**

Vitamin B in high doses can significantly reduce oxalate levels. However, research has shown that this treatment only works in 30-40% of patients with Primary Hyperoxaluric Type 1 (not type 2/3).

**What can I do to help?**

The single most important thing patients can do to help themselves is to drink a lot of fluids (3.5-4.0 litres a day). The fluid does not need to be made up of entirely water. Fruit juices, tea, coffee, etc. all count towards fluid intake. Citrus fruit juices have the additional benefit of alkalinising the urine (patients should be aware of the sugar content).

Patients should try to reduce the amount of salt in their diet. Salt can increase the amount of calcium in the urine and therefore increase the risk of forming a stone with the oxalate. Patients should try not to eat more than 6g salt a day (about the size of a small teaspoon). Speak to your nurse, doctor, or dietician if you would like further information on how to reduce salt in your diet.

Oxalates are present in many foods that we eat. Patients with primary hyperoxaluria may not find much benefit in trying to remove oxalates from their diet all together. This is because the body will continue to generate high oxalate regardless of dietary restriction. However we do advise patients to avoid foods that are severely high in oxalate such as rhubarb, spinach, almonds, beetroot, sesame seeds, and Swiss chard.
Common investigations

Ultrasound scan

An ultrasound scan can be requested to check for oxalate stones within the kidney, or can be used to monitor existing kidney stones. The procedure is safe and painless. You will lie down and a probe will be placed on your skin. The probe sends out pulses of ultrasound which will detect your kidneys and any stones.

X-Ray

An X-ray is safe and painless, and is able to produce images of the entire urinary tract.

CT scan

A CT scan uses a series of X-rays taken at different angles to build a detailed picture of inside the body. They involve a significant dose of radiation and are therefore not used for routine monitoring.

Blood and urine tests

- You will have frequent blood tests to monitor your kidney function, and to monitor the level of waste products and minerals within your blood
- You will have frequent urine tests to monitor for signs of infection. Increased levels of blood or protein in your urine may be an early indication of a stone being formed
- You will be asked to perform regular 24 hour urine collections to monitor your oxalate excretion
- If your kidney function is poor or you are on dialysis treatment, you will have a regular blood test to monitor the amount of oxalate in your blood. This is called a plasma oxalate level and needs to be sent to a special laboratory. Therefore results can take between 2-4 weeks to come back
What should I do if I get renal colic?

Renal colic can be extremely painful and distressing. Small stones can often be passed (up to approx. 5mm) but can still be incredibly painful, although this is different for each patient. If you are in severe pain seek medical advice immediately. You can try and encourage the stone to be passed more quickly by increasing your fluid intake. Painkillers may help take away, or reduce, the amount of pain. Some people find that applying heat to the area can help (warm baths, heat pads etc.).

If you are having pain when passing urine you should have your urine checked for infection. Sometimes urine infections can cause pain in the same areas as renal colic.

You should seek medical advice if you have any of the following symptoms alongside renal colic:

- Fever, nausea, and vomiting
- No longer passing any urine
- Severe pain that cannot be controlled with medication
- Single functioning kidney

Will primary hyperoxaluria affect my family?

Primary hyperoxaluria is inherited in an autosomal recessive pattern. This means that a person must inherit two changed copies of the same gene in order to have primary hyperoxaluria. If a person inherits one changed gene and one normal gene, then that person will be a healthy carrier. If both parents are carriers of the same changed gene, they may pass on either their normal gene or their changed gene to their child.

Each child of parents who both carry the primary hyperoxaluria gene therefore has a 25% chance of inheriting a changed gene from both parents and being affected by primary hyperoxaluria.

There is a 50% chance that the child will inherit just one copy of the primary hyperoxaluria gene. This means they will be healthy carriers like their parents.

There is a 25% chance that the child will inherit both normal copies
of the gene and will therefore not have primary hyperoxaluria or be a carrier of primary hyperoxaluria.

The chance remains the same in every pregnancy and is the same for boys and girls.

### Autosomal recessive inheritance: both parents carriers

Patients with primary hyperoxaluria are advised to discuss any plans for pregnancy with their specialist team.

### Useful resources of information

**The Oxalosis & Hyperoxaluria Foundation:**

The Oxalosis & Hyperoxaluria Foundation are an international organisation based in New York that promotes research into finding a cure and aims to improve the care and treatment for those affected by primary hyperoxaluria. Their website has lots of information about primary hyperoxaluria and has links to other useful websites.

[www.ohf.org](http://www.ohf.org)

**Rare Kidney Stone Consortium**

The Consortium provides support for clinical studies and facilitates the exchange of information and resources among investigators, clinicians, and patients, and researchers in order to improve care and outcomes for patients with rare stone diseases. Their website has information about primary hyperoxaluria and also has updates on ongoing research.

[www.rarekidneystones.org](http://www.rarekidneystones.org)
**Rare Renal**

This website is UK initiative supported by British Kidney Patient Association and Kidney Research UK. The website has lots of information about rare kidney diseases, including primary hyperoxaluria for patients.

www.rarerenal.org

**Advice for when you attend clinic**

There is a specialist kidney clinic for patients with primary hyperoxaluria where you will be seen by a doctor or specialist nurse knowledgeable of primary hyperoxaluria. Below are a few tips you can follow to make the most of your appointment:

- Write down any questions you may have because it can be easy to forget when you enter the busy clinic environment
- Write down any symptoms you may have experienced since your last clinic appointment. Make sure you include when the symptoms started and what makes them worse or better
- Never be afraid to double check and ask for things to be explained if you do not understand
- Ask for any words you do not understand to be written down and explained
- Write down a summary of any plans that have been made or any tests that have been booked
- Ask when your next clinic appointment will be

**MyHealth@QEHB**

MyHealth@QEHB is a secure internet based programme that allows you to view blood results, clinic letters, and much more. Speak to your nurse or doctor if you would like to find out more.

**Help with health care costs**

Information can be found in the leaflet HC11, “Are you entitled to help with health costs?” available from the cashiers office in the Queen Elizabeth Hospital, Birmingham.

Or for more information please visit www.nhs.uk/healthcosts
Useful contacts

Renal Metabolic Disease Nurse Specialist
Tel: 07810 654 864    Office: 0121 371 8708

Renal Outpatients
Nurses: 0121 371 5635 or 0121 371 5633
Appointments: 0121 371 4447 or 0121 371 4446

The Trust provides free monthly health talks on a variety of medical conditions and treatments. For more information visit www.uhb.nhs.uk/health-talks.htm

Renal Department
Queen Elizabeth Hospital Birmingham
Mindelsohn Way, Edgbaston, Birmingham B15 2GW
Telephone: 0121 627 2000