



Information prescription for patients with cystinosis

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We would like to thank Orphan Europe (UK) Limited for kindly paying for the printing of this patient information leaflet.

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Information prescription for patients with cystinosis

What is an information prescription?

Information prescriptions contain information about the condition, treatments, care services, benefits advice, and support groups.

Information prescriptions let people know where to get advice, where to find support, and how to make contact with others with the same condition. They include helpful addresses, telephone numbers, and websites.

The aim is to provide you with information that is specific, comprehensible and timely. It is hoped that this will reduce unnecessary stress, anxiety or confusion about your condition.

The information will allow you to make informed choices about what decisions to take in relation to your health and well-being. The information prescription may make you better informed about your condition which will enable you to voice your needs to healthcare professionals, understanding your condition can also lead to better health outcomes.

How does it work?

You can have as much or as little information as you would like. The information will be given to you in an ongoing basis tailored to your individual needs. You will have the chance to discuss any of the information that has been given to you with a doctor or nurse specialist.

Contents

This information prescription is for patients with cystinosis. The following areas are available for your information prescription:

- Cystinosis: An introduction to cystinosis for patients and families
- Cystinosis: Management and treatment
- Cystinosis and your eyes
- Genetics: Information for patients and families
- Transition
- Young people: Health and personal
- Sexual health and fertility
- Diet, exercise and lifestyle
- Supportive services
- Welfare and financial information
- Helpful contacts
- Travelling with cystinosis
- 'My Care'
- 'My Clinic'

This list is not exclusive. Further information will be developed on an ongoing basis to meet the needs of the patient. Please feel free to add in any other information that you have found that you think is useful.

If you would like to provide feedback or have any suggestions please email renal.metabolic@uhb.nhs.uk.

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Cystinosis: An overview for patients and families

Cystinosis: An overview for patients and families

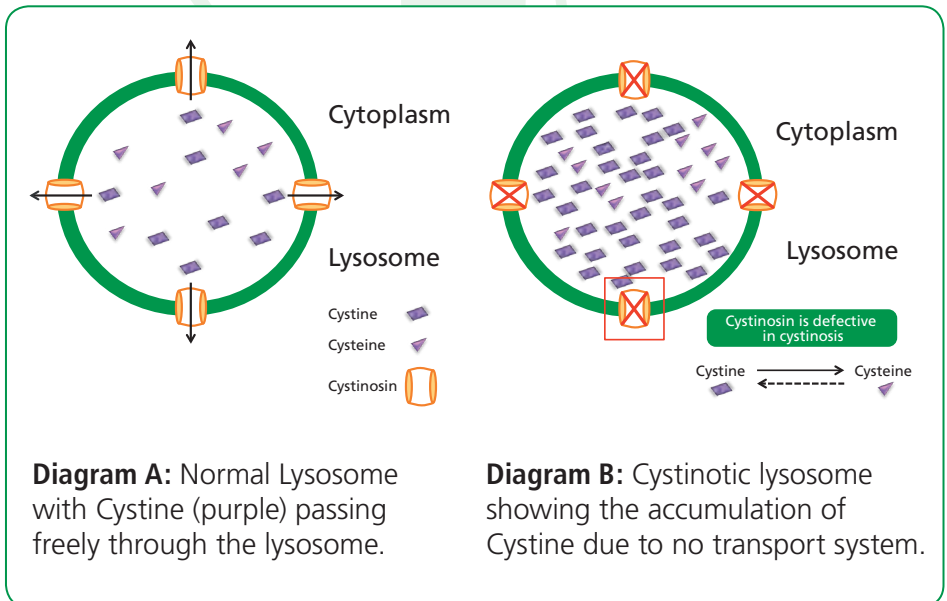
Introduction

Cystinosis is a rare inherited disorder which is estimated to affect 1 in 100,000 to 200,000 people. Cystinosis is characterised by the accumulation of an amino acid called cystine within the lysosomes of the body. Lysosomes are a compartment within the cell that digest and recycle materials such as proteins.

Cystine is an amino acid which is a building block of protein and is essential to life. Protein is broken down inside the lysosomes into several amino acids (including Cystine). In a patient without cystinosis cystine is allowed to leave the lysosome to be used again by the body.

In patients living with cystinosis there is no transport system available to carry cystine out of the lysosomes therefore causing cystine to accumulate within the cell.

The accumulating cystine will eventually form into crystals within your cells and start to affect various organs such as the kidneys, eyes, muscle, pancreas and the brain.



Types of cystinosis

Early onset: *Infantile cystinosis*

This is the most common form of cystinosis. The first symptoms generally appear several months after birth. Without treatment renal failure may develop between the age of 8 and 12 years.

Symptoms may occur within the first year or two of life. These include:

- Poor growth and development
- Poor appetite
- Excessive thirst
- Excessive urination
- Dehydration
- Ricketts

These symptoms are caused by the damage done to the kidneys. The kidney is unable to concentrate the urine and allows important electrolytes such as sodium, potassium, phosphorus and bicarbonate to be wasted into the urine. This is often referred to as Fanconi Syndrome.

Cystine crystals in the eye can be seen with a slit lamp eye test (a small strip of light shone in the eye) between 12 and 16 months of age. This can lead to pain and discomfort from bright lights (photophobia).

Late onset: *Juvenile cystinosis*

This is a rare form of cystinosis. The symptoms are comparable to those of infantile cystinosis but with later onset, usually in adolescence or early adult life. Renal failure can develop between the ages of 20 and 30 years.

Non-nephropathic cystinosis

This form is sometimes described as ocular cystinosis. It is usually discovered at adult age through performing other routine

examinations such as an eye test. Patients with this form rarely report any symptoms and damage to other organs generally does not occur.

<p>Child (Infantile cystinosis)</p> <ul style="list-style-type: none">• Failure to thrive• Photophobia• Ricketts• Vomiting• Dehydration• Excessive urination• Developmental delay• Corneal crystals• Kidney damage from the age of 2• Low potassium• Low phosphate• Too much acid in the blood	<p>Late onset cystinosis</p> <ul style="list-style-type: none">• Symptoms same as infantile• Presents from the age of 10 up to early adulthood• Will reach end stage renal failure between 15 and 25 years of age	<p>Adult (Benign cystinosis)</p> <ul style="list-style-type: none">• Corneal crystals• Photophobia• No other organ damage
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Treatment

Symptomatic treatment (before kidney failure develops)

It is possible to relieve some of the kidney symptoms by ensuring that you drink an adequate amount of water to replace the excessive fluid loss.

Taking supplements will replace the sodium, bicarbonate and potassium being leaked by the kidneys. The amount of supplements you take may vary according to your blood results. Vitamin D and phosphorus supplements will heal and prevent bone diseases.

Cystinosis specific treatment

There is currently no complete cure for cystinosis. The aim of specific treatment is to reduce the amount of cystine within the cells.

Cysteamine is a cystine-depleting drug which reduces the level of cystine within the lysosomes.

Cysteamine is most effective when taken four times a day at six hourly intervals. This medication is a life-long treatment and must be taken every day

Cysteamine treatment reduces the progression towards kidney failure. It is not an easy medicine as it has a smell that some people find unpleasant and has to be taken regularly in the long term.

Cysteamine is still effective even after kidney failure has developed. It prevents other organs in the body being damaged and can prevent further damage to the pancreas and liver.

Eye drops containing cysteamine can prevent damage to the eye and can help with photophobia (discomfort in bright light). These need to be applied several times a day to prevent cystine crystals forming.

Treatment monitoring

It is possible to measure the effectiveness of treatment by taking a white blood cell (WBC) cystine level. This is a blood test that allows us

to measure the amount of cystine accumulating in the cells and should be checked every 3-4 months. To prevent damage to your organs the level should be kept below 1.0. Blood samples should be taken 5-6 hours after taking cysteamine. It is important to let your healthcare professional know if you are missing any doses of cysteamine.

How do you get cystinosis?

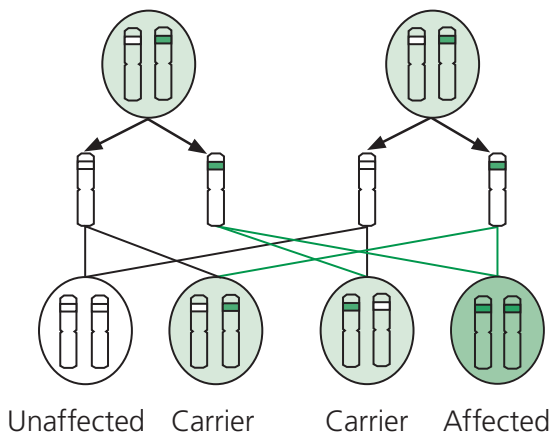
Cystinosis is a genetic inherited disease and is not infectious, contagious or brought on by lifestyle.

We have thousands of genes, each carrying their own set of instructions. When a gene is altered it can result in a genetic condition or disease. We all have two copies of each gene. One inherited from our mother and one inherited from our father.

Cystinosis is inherited in a way that is called recessive. This means that individuals must inherit two copies of the altered gene to be affected by cystinosis. Individuals who inherit one copy of the altered gene are completely healthy and are known as carriers.

When both parents are carriers of the same altered gene there are four possible outcomes for each pregnancy (see diagram below).

Autosomal recessive inheritance: both parents are carriers



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

There is a 50% chance that the child will inherit just one copy of the cystinosis 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 not have cystinosis or be a carrier of cystinosis.

Management of cystinosis

Management of cystinosis

Cysteamine treatment

There is currently no complete cure for cystinosis. The aim of specific treatment is to reduce the amount of cystine within the cells. Cysteamine is a cystine-depleting drug which significantly reduces the level of cystine within the lysosomes.

Cysteamine is most effective when taken four times a day at six hourly intervals. This medication is a life-long treatment and must be taken every day.

Benefits of cysteamine therapy

- Cysteamine treatment has significantly changed the course of the disease
- The progression of renal failure in children treated with cysteamine sufficiently early was delayed from the age of ten to the mid 30s
- Cysteamine treatment also reduces the damage to other organs, in particular the muscles and nervous system
- To achieve the maximum efficacy of cysteamine it is important to start treatment early, to have regular monitoring, adequate dosing, and good compliance

Side effects

It is important to remember that everybody is different and not all patients will have side effects. Unfortunately cysteamine has some potentially unpleasant side effects. These include:

- Vomiting
- Nausea
- Diarrhoea
- Lethargy

- Skin and breath odour

Cysteamine may cause drowsiness. When you start therapy or have a dose increased you should not engage in potentially hazardous activities (e.g. driving) until you are aware of the effects.

If you are experiencing any side effects or would like further information about side effects please discuss this with a healthcare professional.

Taking cysteamine

Due to the unpleasant side effects it is understandable that taking cysteamine can sometimes be difficult.

It is important that you discuss any concerns you have about your medication with a healthcare professional.

Do not be scared to talk to your hospital team if you are not managing to take your cysteamine as there are often ways to remedy unpleasant side effects.

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Common medications

Common medications

The following is a brief guide to a few of the medications you may be prescribed. Medications are prescribed depending on the need of the individual therefore you should not be alarmed if you are not on all of these. Equally you may be prescribed additional medications that are not listed. When you are prescribed a new medication we encourage you to discuss why you need the medication and its side effects with your hospital team.

Do not be alarmed when reading the side effects. Medications can affect people differently and it's rare that anybody experiences all of the side effects. It may be that you only experience one or two mild effects, or you may not even experience any at all.

Cystinosis specific medication

Medication: Cysteamine

Brand names: Cystagon® (white capsule)

Why do I need it? Cysteamine severely reduces the amount of cystine within the cells. If started early enough delay the progression of kidney failure and reduce the damage done to other organs.

Possible side effects: Side effects include vomiting, nausea, diarrhoea, lethargy, skin odour and breath odour. If you would like to know more about the side effects please discuss this with a healthcare professional.

Medication: Cysteamine eye drops

Brand names: Available in varying strengths – 0.55%, 0.11%

Why do I need it? Cystine crystals can deposit in the cornea. This can feel uncomfortable and make you sensitive to bright lights. If left untreated it can affect your vision. Eye drops are the only way to dissolve the cystine crystals in your eye.

Possible side effects: Application may slightly sting when you first start using the eye drops.

Medication: Levothyroxine

Brand names: Eltroxin® 50mcg, 100mcg (round, white/off white colour)
Evotrox® 50mcg, 100mcg (liquid solution)

Why do I need it? Cystinosis can cause the thyroid gland to not produce enough thyroid hormone (hypothyroidism) which can cause weight gain, depression, joint pain, muscle cramps, and low heart rate. This can be treated by taking Levothyroxine.

Possible side effects: Side effects can include diarrhoea, vomiting, headaches, palpitations and restlessness.

Carnitine

Brand names: Carnitor® (oral liquid)

Why do I need it? Carnitine is used by your body to help make energy from fat. Patients with cystinosis have low Carnitine levels because it is lost in the urine.

Possible side effects: Nausea, vomiting, abdominal pain and diarrhoea. These side effects can be related to the dose you are taking. Your tolerance should be monitored during the first week and after any dose increase.

Proton pump inhibitors (PPI)

Lansoprazole, Omeprazole (capsule)

PPIs can help reduce some of the gastro-intestinal discomfort that is associated with taking cysteamine tablets.

They are generally well tolerated but some patients can experience nausea, abdominal pain, diarrhoea, flatulence and constipation.

Supplements (Various)

Brand names: Potassium, Sodium, Bicarbonate, Phosphate, Vitamin D

Why do I need it? You may need to take additional supplements to replace any important electrolytes or vitamins that your kidney wastes. The type of supplements and the dose you take will depend on your blood results.

Possible side effects: Side effects can vary greatly between patients. If you feel like you are experiencing any side effects when taking supplements you should speak to a healthcare professional.

Medication: Insulin replacement

Why might I need it? Patients with cystinosis are at an increased risk of getting diabetes. There are many different types of insulin replacement treatments available which should be discussed with your diabetic team.

Transplant specific medication

If you have received a kidney transplant you may come across the following medication. If you would like further information about transplantation we encourage you to speak to your local transplant team.

Medication: Tacrolimus

Brand names: Prograf®, Advagraf®

Why do I need it? If you've had a transplant this medication stops the body attacking the new kidney. Seek further advice from your transplant team.

Possible side effects: Side effects can include tremors, nausea, blurred vision, sleep problems. This medication will also reduce your natural defences against infection. Seek advice from your transplant team if you are experiencing any problems.

Medication: Cyclosporine

Brand names: Neoral®, Sandimmun®, Deximune®

Why do I need it? This is another medication used to stop the body attacking a transplanted kidney. Seek further advice from your transplant team.

Possible side effects: Side effects include tremors, nausea, increased hair growth, swollen gums, and numbness or tingling in your feet, hands or face. This medication will also reduce your natural defences against infection. Seek advice from your transplant team if you are experiencing any problems.

Medication: Azathioprine

Azathioprine, Imuran®

Why do I need it? This reduces the amount of white blood cells that can attack the transplanted kidney.

Possible side effects: Side effects may include nausea, vomiting, diarrhoea, fever and rash. This medication will reduce your natural defences against infection. Seek advice from your transplant team if you are experiencing any problems.

****IMPORTANT****

If you have any concerns with your medication it is important to discuss this with a healthcare professional. If you are regularly missing doses you need to inform your doctor.



Common investigations

Common investigations

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.

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Cystinosis and your body

Cystinosis and your body

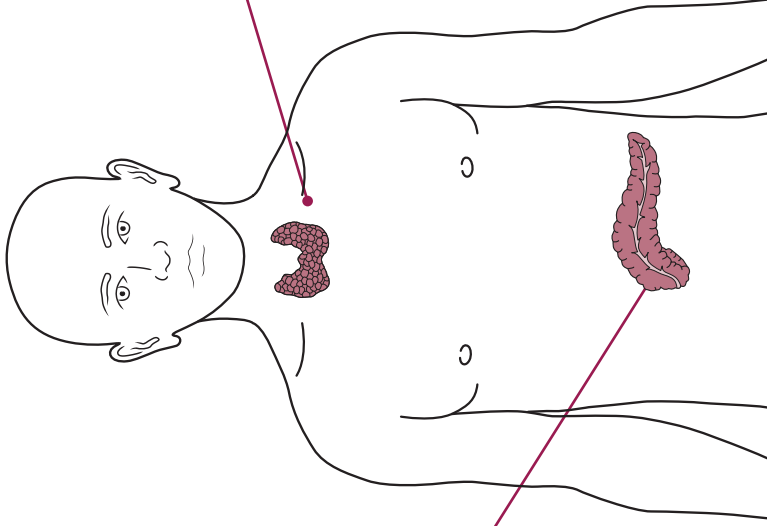
Cystinosis can potentially affect many parts of your body, not just your kidney. It is important that you are aware of potential complications so that treatment can be started as soon as possible.

Diabetes

In later life patients with cystinosis are at an increased risk of having diabetes. Cystine crystals deposited in the pancreas can cause the body to slowly stop producing insulin. Eventually it will be necessary to start insulin replacement therapy.

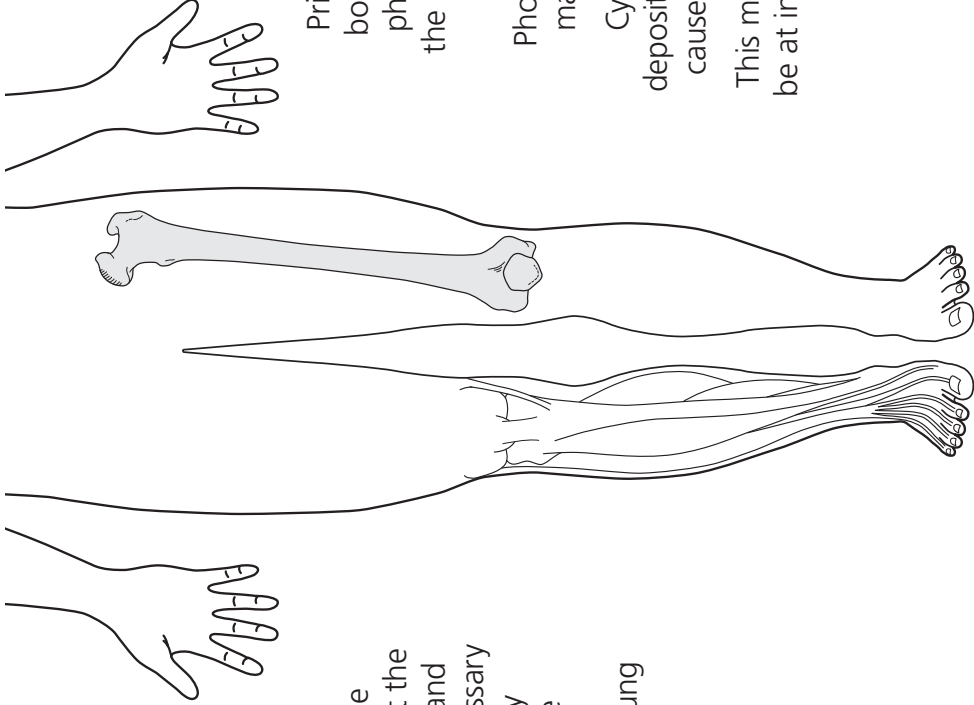
Thyroid

The accumulation of cystine in the thyroid gland causes the thyroid gland to not function properly. This can be diagnosed through regularly blood checks and is easily treatable by taking a supplementary tablet called Levothyroxine.



Muscles

Cystine crystals deposited in the muscles can lead to muscle weakness. This can also affect the muscles used for swallowing and breathing. It is therefore necessary to monitor your swallowing by seeing a speech and language therapist. Your chest muscles can be monitored by having lung function tests.



Bones

Prior to having a transplant bones can be weakened by phosphate being wasted in the urine due to early kidney damage.

Phosphates are important in maintaining healthy bones.

Cystine crystals can also be deposited in the bones and can cause metabolic bone disease.

This means that the bones may be at increased risk of fractures.

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Cystinosis: Looking after your eyes

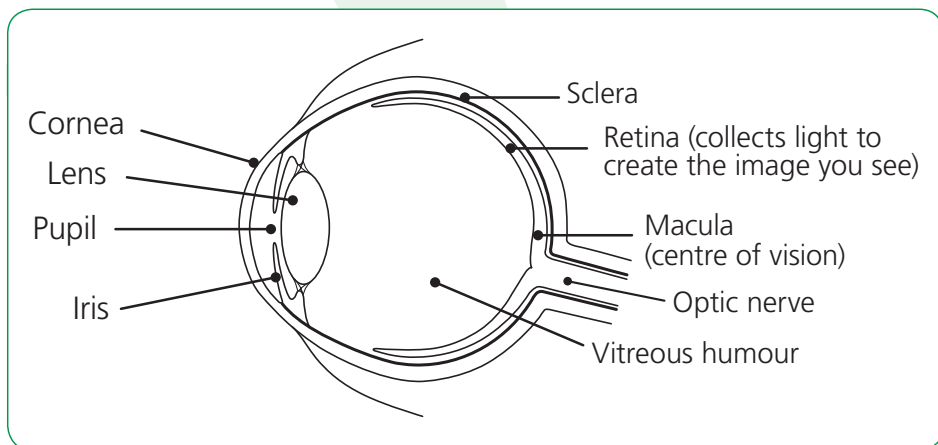
Cystinosis: Looking after your eyes

How does cystinosis affect my eyes?

Cystinosis causes cystine crystals to be deposited in the cornea of the eye. The cornea is the transparent front part of the eye that covers the iris and the pupil.

Cystine crystals in the cornea cause light to be reflected which can result in substantial discomfort to bright light (photophobia).

If left untreated excessive corneal crystals can give you a gritty feeling in your eye which may cause you to experience some pain. Untreated corneal crystals also affect your visual acuity which means your vision may appear less clear and less sharp.



What treatment is available?

Unfortunately oral cysteamine has no affect on corneal crystals. Therefore cysteamine eye drops need to be applied to dissolve the cystine crystals.

Cysteamine eye drops will relieve the discomfort from bright lights and start dissolving the crystals at any age.

Cysteamine eye drops should be applied regularly every day. Eye drops should be applied every couple of hours. Compliance can be difficult but cysteamine eye drops are currently the only way to

reverse corneal crystals and prevent any future damage to the eye.

Attending an optician about corneal crystals is not sufficient. You should be reviewed regularly by an ophthalmologist who recognises and understands cystinosis.

Cystinosis: Genetics and inheritance

Cystinosis: Genetics and inheritance

Introduction to genes and chromosomes

Your body is made up of millions of cells, most of which contain a complete set of genes.

Genes act like a set of instructions for our bodies to follow, controlling our growth and how our bodies work. They are also responsible for characteristics such as eye colour, blood type and height.

We inherit two copies of each gene, one copy from our mother and one copy from our father. We all have thousands of genes in total.

Our genes are located on chromosomes. We have 46 chromosomes (23 pairs). We inherit 23 chromosomes from each of our parents.

Sometimes there is a change in one of the genes which stops the body from working properly.

How is cystinosis inherited?

Cystinosis 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 cystinosis. 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.

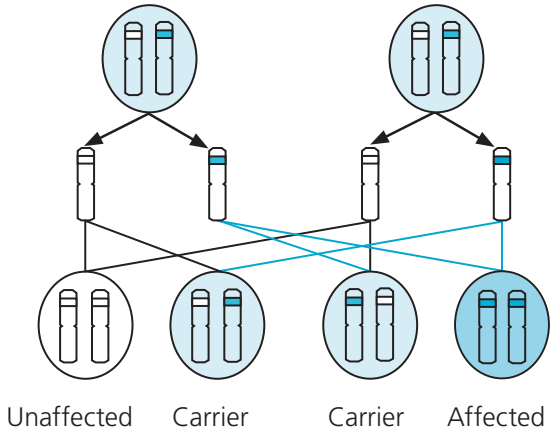
Each child of parents who both carry the cystinosis gene has a 25% chance of inheriting a changed gene from both parents and being affected by cystinosis.

There is a 50% chance that the child will inherit just one copy of the cystinosis 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 not have cystinosis or be a carrier of cystinosis.

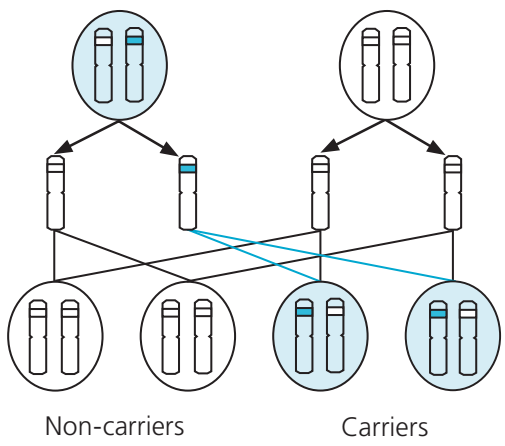
The chance remains the same in every pregnancy and is the same for boys and girls.

Autosomal recessive inheritance: both parents are carriers



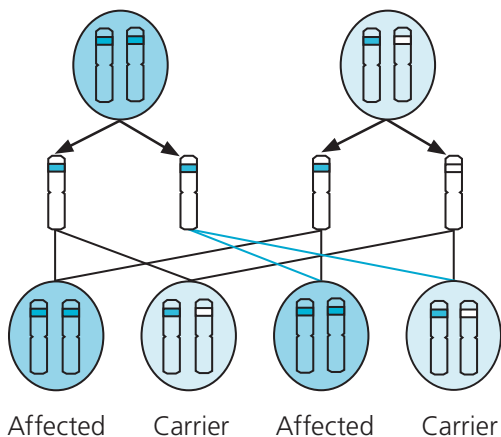
If only one parent is a carrier of the cystinosis gene, each child will have a 50% chance of being a carrier but will not be affected by the condition.

Autosomal recessive inheritance: one parent is a carrier



If one parent is affected and the other is a carrier, then there is a 50% chance the child will be affected by cystinosis. There is 50% chance the child will be a carrier of the cystinosis gene and be unaffected.

Autosomal recessive inheritance: one parent affected one parent carrier



If one parent has cystinosis and the other is not a carrier there is 100% chance the child will be a carrier of the cystinosis gene but will not be affected by the condition.

Carrier testing in pregnancy

If both parents are known to be carriers of the cystinosis gene, and are considering having a baby, prenatal detection may be an option to see if the baby has inherited cystinosis.

Chorionic villus sampling involves taking a tiny sample of tissue from the developing placenta and is usually performed at 10-13 weeks.

Amniocentesis is when a sample of fluid is taken from the amniotic sac and can be performed after 15 weeks.

The discussion to have prenatal testing can be very difficult and you should only make a decision following consultation with your genetic councillor/genetic consultant and if you, and your partner, feel that it is important to know the information that the prenatal test will give.

www.geneticalliance.org.uk and **www.nhs.uk** has further information about prenatal genetic testing.

Contacts

Genetic Alliance UK

Telephone: 02077 043 141

Email: contactus@geneticalliance.org.uk

Website: www.geneticalliance.org.uk

A directory of regional genetic centres in the UK can be found at
www.geneticalliance.org.uk/services.htm

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Transition from paediatric care

Transition from paediatric care

Transition between being a teenager and adulthood can present some very difficult challenges. Young adults with cystinosis have some unique challenges in addition to the usual challenges that teenagers have to deal with.

This information aims to briefly explain transition and what you should expect when transferring to adult services.

What's different about adult services?

The main difference is that the environment may appear more formal and clinic will be run on a lot larger scale. Initially it may seem overwhelming but try not to let this deter you and ensure you voice any concerns.

You will have more responsibility for your health and the care you receive; therefore decisions will likely be made between you and the doctor.

How do I make transition easier?

Transition is best when it is personalised to you and your family. The healthcare professional will work with you and your family as a team to ensure a healthy independence is achieved. The following tips may be useful to you but remember that everyone is individual so do not feel under pressure to achieve everything straight away.

- Ask your doctor if your hospital has a transition programme in place
- Arrange a visit to the Adult hospital to familiarise yourself with the surroundings. This will also give you the opportunity to meet the staff that will take over your care
- Try to get involved in any healthcare related discussions about your care and treatment choices.
- Take an interest in monitoring your condition. Try checking your blood results on [Renal Patient View](#)

- Try taking some responsibility over your medications. Start learning what each one does and the side effects. Speak to your doctor about any problems you're having with your medication
- Keep a record of your clinic appointments
- Find out how to make your way to clinic on your own
- Make sure you know who to contact in an emergency
- Don't be afraid to talk to your friends and family about how you are feeling. This may seem hard at first but sharing your problems may make them easier to cope with
- Try making contact with other patients with cystinosis to discuss how they felt during transition. If you don't know anyone try asking your healthcare professional to put you in contact with another patient

Transition is not restricted to issues you have with cystinosis. Try and take the opportunity to discuss sex and sexually transmitted diseases, birth control, alcohol, and drugs.

Knowledge is power!

Being knowledgeable about cystinosis and learning to take control of your condition can aid you in a successful transition. It is well documented that being knowledgeable about your condition results in better outcomes and an increased quality of life.

Speak to your healthcare professional if you would like to learn more about cystinosis. They may be able to arrange some informal teaching or point you in the right direction of reliable information.

If you want you can use the following tool to check your progression through transition.

Questions	✓	Comments
I can briefly explain what cystinosis is and what affect it has on my health.		
I am knowledgeable about my medical history.		
I have been to the adult hospital and met the staff.		
I can access Renal Patient View and monitor my blood results.		
I know what medications I take, what they are for and the implications of not taking them.		
I ask questions in clinic.		
I take part in discussions about my treatment.		
I have had the opportunity to discuss and ask questions about alcohol and drugs.		
I have had the opportunity to discuss and ask questions about sex, sexuality, sexually transmitted diseases and birth control.		
I know who to contact in an emergency.		
I am aware of relevant support groups and local supportive services.		
I know how to get to and from clinic on my own.		
I keep a record of future appointments and know who to contact if I can't attend.		
I have discussed my health aims and goals with my healthcare professional.		

Remember that transition is a very personal and gradual process. If you feel like things are moving too fast or you are not coping it is important to discuss this with your healthcare professional and family.

Useful contacts

www.nhs.uk/livewell/sexandyoungpeople

www.talktofrank.com

www.brook.org.uk

www.kidneymatters.co.uk

www.britishkidney-pa.co.uk

www.kidneypatientguide.org.uk

www.cystinosis.org.uk

www.cystinosis.com

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Young people with cystinosis

Young people with cystinosis

Living with a chronic condition

Transition from childhood to adulthood can be a particularly tough time and having a chronic condition, such as cystinosis, doesn't make this transition any easier.

It's not abnormal to feel like you are not coping. Try to talk about how you feel with your family and friends. It's a good idea to be aware of the signs of depression as this is entirely treatable. The signs of depression include:

- Feeling sad
- Getting angry easily
- Not eating at all or eating too much
- Feeling tired
- Feeling anxious
- Sleeping too much or too little
- Thinking about self harm

You should not feel embarrassed or scared to discuss this with people close to you. Informing your healthcare team to seek help is essential.

The smell of Cystagon

Cystagon is currently the only treatment for cystinosis and the only way to minimise damage to your organs. Taking your Cystagon decreases and delays the onset of complications and improves life expectancy.

Unfortunately Cystagon can lead to some unpleasant side effects. Gastrointestinal, unpleasant body and breath odour in particular are difficult for young people to cope with. At an age where you are looking to build new relationships and undertake social activities it is understandable that it may be tempting to skip doses or stop all together. However this is not advisable as skipping doses can be

detrimental to your health and can damage your organs without you realising.

You should discuss any issues you are having with side effects with your healthcare team. There are often ways to deal with the odour of Cystagon such as Vitamin B, body sprays and breath sprays.

Getting to know your cystinosis

It's important for you to be able to answer questions about your health. In an emergency situation it would be useful to know:

- When you received your transplant or when you started renal replacement therapy
- What medication you take? When you take them? And why do you take them?
- Are you allergic to anything?
- Recent illness, infection or admission to hospital
- Your history of surgery and any other medical problems you may have
- Who your main doctors are and how to get hold of them

Knowing this information will help the doctors make an accurate diagnosis in an emergency.

Networking

Ask your healthcare team if they can put you in touch with another patient who has cystinosis. Some people find it useful to discuss their problems and difficulties with people in a similar situation.

Make contact with The Cystinosis Foundation UK. They will be able to provide you with information, support and keep you updated on any future events.

Moving out

The law does not allow people under the age of 18 to have a mortgage or hold a tenancy on a rented property.

However a tenancy on a rented property can be granted if you have a joint tenant who is 18 or over.

To apply for council housing you should apply through your local council. You usually need to be 18 to apply for council housing. However some councils will let you join the waiting list at 16. Council housing is usually offered to those who need it most. For further information you should visit:

- Your local council website
- **www.gov.uk**

Employment

Throughout your working life you may experience concerns and issues that are unique to an individual with cystinosis. However living with cystinosis should not limit your choice of employment. Your choice of career should be entirely based on your intellect, ability, interests and life goals.

You should never be treated less favourably because you have cystinosis. An employer has a duty to make 'reasonable adjustments' to avoid you being disadvantaged compared to other employees. 'Reasonable adjustments' can include providing you with adapted equipment or adjusting your working hours.

If you feel like you are being discriminated against it is advised you discuss this with your employer and inform them of your rights and their responsibilities under the Equality Act 2010. If you are not satisfied you can make a complaint to the employment tribunal. All complaints must be made within three months of the alleged discrimination.

Further information about the Equality Act 2010 and your rights is available at: **www.gov.uk/discrimination-your-rights**

Equality and Human rights Commission: www.equalityhumanrights.com

Citizens Advice Bureau: www.adviceguide.org.uk

Employment Tribunal (general enquiry line): 0845 795 9775



**Cystinosis sexual health and
fertility**

Cystinosis: sexual health and fertility

Fertility in men

Puberty in male patients can be delayed by 2-3 years. The accumulation of cystine in the testes causes a decrease in the amount of testosterone produced by the testes. Male patients with nephropathic cystinosis reach puberty at approximately 16-17 years of age.

The ability to have an erection and engage in a sexual relationship is unaffected.

Infertility is very common amongst males with nephropathic cystinosis and there have been no reported cases of a male with nephropathic cystinosis fathering a child. Research has shown that patients with nephropathic cystinosis, even with early cysteamine treatment, produce no, or very little, sperm. Although it has been suggested that in vitro fertilisation may be possible for some patients.

Fertility in women

Female patients with cystinosis are generally believed to reach puberty at approximately 14-15 years of age. Ovulatory cycle development and sexual development are relatively normal in female patients.

Female patients have a good chance of giving birth to a child. There have been several reported cases of women with cystinosis giving birth to a healthy baby, providing renal/transplant function is satisfactory.

However no one is sure what affect cysteamine has on the developing foetus. It is therefore very important that if you are planning to start a family, or conceive unexpectedly, you discuss this with your consultant.

Contraception

Using contraception is the use of hormones, devices, or surgery to prevent a woman from becoming pregnant. It is important to know that most types of contraception do not protect against sexually transmitted infections. The male condom is the only form of contraception that protects against STIs as well as pregnancy.

Find out about all the different types of contraception at www.nhs.uk/conditions/contraception. Contraception is available through your GP, community contraceptive clinics, sexual health clinics and some young people's services.

If you have had a transplant it is important that you discuss contraception with your transplant team as some types of contraception may not be suitable.

Glossary

Azoospermia: A condition in which the male has no measurable level of sperm in the semen.

Foetus: An unborn baby more than 8 weeks after conception.

In Vitro Fertilisation: The process whereby an egg is fertilised with sperm outside of the body and the resulting embryo is transplanted into the women's uterus.

Ovulatory: In relation to ovulation; the release of the ovum (the egg) from the ovary.

Sperm: The male reproductive cell.

Semen: The male reproductive fluid, containing sperm.

Testosterone: A hormone produced mainly by the testes that stimulates male secondary sexual characteristics.

Useful article:

Besouw, M. et al: Fertility Status in Male Cystinosis Patients Treated with Cysteamine.



Supportive services

Supportive services

People living with a rare condition can sometimes feel isolated and frustrated. However you are not alone and there are charities and support groups who all offer support and advice to patients and families.

Cystinosis Foundation UK

The Cystinosis Foundation UK registered as a charity in 1999 and has continued to provide support to all people diagnosed with cystinosis, their families and their relatives. The foundation works with cystinosis-related charities worldwide to help co-ordinate research support and maintain awareness of new treatments.

Website: www.cystinosis.org.uk

Cystinosis Research Network

The Cystinosis Research Network are based in the USA and are dedicated to supporting and advocating research, providing family assistance and educating the public and medical communities about cystinosis.

Website: www.cystinosis.org

Genetic Alliance UK

Genetic Alliance UK aim to improve the lives of people affected by genetic conditions and to raise awareness and improve information for patients and families. They provide a united voice for all those affected by a genetic condition and actively campaign on issues of policy and practice.

Website: www.geneticalliance.org.uk

Children Living with Inherited Metabolic Disorders (Climb)

Climb is a national organisation who works on behalf of children, young people, adults and families affected by metabolic disease. Climb is the UK's foremost provider of free metabolic disease information. Their vision is to provide support and information to children, young people, adults and families.

Website: www.climb.org.uk

Rare Disease UK

RDUK is the national alliance for people with rare diseases and all who support them. RDUK is campaigning for health departments across the UK to develop a strategy for rare diseases to ensure patients living with rare conditions have equitable access to effective services.

Website: www.raredisease.org.uk

British Kidney Patients Association (BKPA)

The BKPA is a well established charity that works to improve the quality of life for adults and children with kidney disease. They can offer information and advice to patients and families. They also award grants to patients and families to help with domestic bills, education and holidays.

Website: www.britishkidney-pa.co.uk

The National Kidney Federation

The NKF aims to promote best renal practice and treatment, and the health of those suffering from chronic kidney disease or who establish renal failure. The NKF supports the related needs of relatives and friends who care for kidney patients.

Website: www.kidney.org.uk

Kidney Research UK

Kidney Research UK campaign for better patient access to renal services and are dedicated to raising awareness of kidney disease. They award grants to kidney patients so that they can cope with the unexpected costs of their condition. They also operate a kidney information service to answer any questions patients may have about their condition.

Website: www.kidneyresearchuk.org

Helpful contacts

Cystinosis Foundation

Email: Available through 'contact us' section of website

Address: Cystinosis Foundation UK
174 Corwen Road
Tilehurst
Reading
RG30 4TA

Rare Disease UK

Telephone: (020) 7704 3141

Email: info@raredisease.org.uk

Address: Genetic Alliance UK
Unit 4D, Leroy House
436 Essex Road
London
N1 3QP

British Kidney Patients Association

Telephone: 01420 541 424

Email: info@britishkidney-pa.co.uk

Address: The BKPA
3 The Windmills
St Mary's Close, Turk Street
Alton
GU34 1EF

The National Kidney Federation

Telephone: (01909) 544 999

Email: nkf@kidney.org.uk

Address: The NKF
The Point, Coach Road
Shireoaks
Worksop
S81 8BW

Climb

Telephone: 0845 241 2173

Email: enquiries@climb.org.uk

Address: Climb Building
176 Nantwich Road
Crewe
CW2 6BG

Genetic Alliance

Telephone: (020) 7704 3141

Email: Contactus@geneticalliance.org.uk

Address: Genetic Alliance UK
Unit 4D, Leroy House
436 Essex Road
London
N1 3QP

Rare Disease UK

Telephone: (020) 7704 3141

Email: info@raredisease.org.uk

Address: Genetic Alliance UK
Unit 4D, Leroy House
436 Essex Road
London
N1 3QP

Kidney Research UK

Telephone: 0845 070 7601

Email: enquiries@kidneyresearchuk.org

Address: Enquiries Kidney Research UK
Nene Hall
Lynch Wood Park
Peterborough
PE2 6FZ

Welfare and supportive services

Welfare and financial services

It is important to get all the help that you are entitled to. The area of welfare and benefits can sometimes appear quite daunting for people. The following information aims to provide you with useful information and helpful contact information.

Prescription charges

Patients living in England currently have to pay for prescriptions. Patients living in Northern Ireland, Scotland and Wales receive free prescriptions. However there are certain medications that are free of charge and patients with certain medical conditions are exempt from paying prescription charges.

You can get free NHS prescriptions if, at the time the prescription is dispensed, you:

- Are 60 or over
- Are under 16
- Are 16-18 and in full-time education
- Are pregnant or have had a baby in the previous 12 months and have a valid maternity exemption certificate (MatEx)
- Have a specified medical condition and have a valid medical exemption certificate (MedEx)
- Have a continuing physical disability that prevents you from going out without help from another person and have a valid MedEx

You are also entitled to free prescriptions if you or your partner (including civil partners) are named on, or are entitled to, an NHS tax credit exemption certificate or a valid HC2 certificate (full help with health costs), or you receive either:

- Income Support
- Income-based Jobseeker's Allowance
- Income-related Employment and Support Allowance, or
- Pension Credit Guarantee Credit

Medical exemption (MedEx) certificates are issued on application to

people who have:

- A permanent fistula (for example caecostomy, colostomy, laryngostomy or ileostomy) requiring an appliance or continuous surgical dressing
- A form of hypoadrenalism (for example Addison's disease) for which specific substitution therapy is needed
- Diabetes insipidus or other forms of hypopituitarism
- Diabetes mellitus, except where treatment is by diet alone
- Hypoparathyroidism
- Myasthenia gravis
- Myxoedema (hypothyroidism requiring thyroid hormone replacement)
- Epilepsy requiring continuous anticonvulsive therapy
- Continuing physical disability that prevents the person from going out without help from another person. Temporary disabilities do not count even if they last for several months

Having cystinosis does not directly entitle you to free prescriptions. However you can claim for conditions on the list that are caused by cystinosis such as diabetes and hypothyroidism. To apply for a MedEx certificate you will need to complete the FP92A form which is available from your doctor.

Prescription prepayment certificates (PPC) are available to help you save money on your prescription charges. There are two PPC options available:

1. A three month PPC costs £29.10 and will save you money if you need four or more items in the three months
2. A 12 month PPC costs £104.00 and will save you money if you need more than 14 items in a year

For information on prescription charges go to:

www.nhs.uk/NHSEngland/Healthcosts/Pages/Prescriptioncosts.aspx

For information on PPCs and to apply online go to:

www.nhs.uk/NHSEngland/Healthcosts/Pages/PPC.aspx

Low income scheme

The scheme provides income related help to people who are not exempt from charges but who may be entitled to full or partial help if they have a low income. The scheme covers NHS prescriptions, NHS dental treatment, sight tests, glasses and contact lenses, and travelling to receive NHS treatment.

For application forms further information on the Low Income Scheme visit www.nhsbsa.nhs.uk.

Citizens Advice Bureau

The Citizens Advice Bureau aims to provide the advice people need for the problems they face. The benefits advice section at www.adviceguide.org.uk provides information on benefits, tax credits, council tax, housing costs, national insurance, and how to appeal or complain about a benefits decision.

What benefits can I get?

The Department of Work and Pensions has made an online guide which you can use to check what you may be eligible to get. It can also give you an estimate on what you may receive.

You can access the guide at www.gov.uk in the 'money, tax and benefits' section.

Useful contacts/further information

Social worker:

Most hospitals have a social worker who should be able to advise you on any issues about benefits and NHS costs.

British Kidney Patient Association (BKPA):

The BKPA offers financial assistance to patients at any stage of treatment if kidney disease is having a serious impact on quality of

life and if patients are struggling on a low income. The BKPA can assist with financial aid, travel costs, further education and training, and holidays.

For further information and guidance on how to apply visit **www.britishkidney-pa.co.uk/grants**.

Kidney Research UK:

Kidney Research UK awards grants to patients who have unforeseen and additional costs as a direct consequence of their kidney condition. Guidance for applying for a grant is available at **www.kidneyresearchuk.org/health/financial-help-for-patients.php**.



**Healthy living for people with
cystinosis**

Healthy living for people with cystinosis

Introduction

Living with a life long condition, such as cystinosis puts you at a higher risk of contracting an illness or getting an infection. Therefore it is important that you lead the healthiest possible lifestyle so that you give yourself the best chance of fighting off an illness.

General advice

There are a number of lifestyle choices everybody should try and make, whether they have a condition or not. These are:

- Not smoking
- Having a healthy balanced diet
- Exercise
- Try to avoid stress
- Drink alcohol in moderation
- Stay warm in the winter
- Use protection against the sun (sun creams etc.)

Ask for advice from a healthcare professional if you would like advice on modifying your lifestyle.

Cystinosis specific advice

- There are some tips specific to cystinosis that you should try and follow, particularly if your kidneys are damaged, if you want to lead a healthy life and protect your kidneys and other organs. These are:
- Take your medication as prescribed and inform a healthcare professional if you are taking any over the counter medication
- Attend your clinic appointments to ensure you're receiving the best and appropriate care

- Follow advice from the healthcare team about diet and fluid intake
- Involve yourself in your care through regular discussions with your healthcare team. Agree on a 'care plan' to ensure all your needs are met
- Keep up to date with the relevant vaccinations
- Seek medical advice if you feel unwell
- Consider taking steps to ensure people are aware of your condition and who to contact in an emergency such as medical alert jewellery

Where can I find more information?

See the below websites for specific information on the kidneys and cystinosis:

- The National Kidney Federation website – www.kidney.org.uk
- Kidney Research UK website – www.kidneyresearchuk.org
- British Kidney Patient Association – www.britishkidney-pa.co.uk
- Cystinosis Foundation UK – www.cystinosis.org.uk
- Rare Renal Diseases – www.rarerenal.org
- Cystinosis Research Network – www.cystinosis.org

The following NHS websites can provide you with a wealth of information about various health topics and point you in the right direction of local services:

- NHS Stop Smoking Services website – www.smokefree.nhs.uk
- NHS Immunisation Information website – www.immunisation.nhs.uk
- NHS Choices – www.nhs.uk
- NHS Live well – www.nhs.uk/livewell
- Change 4 Life – www.nhs.uk/change4life

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Travelling with cystinosis

Travelling with cystinosis

Going on holiday can be a very exciting and enjoyable experience and there is no reason why cystinosis should prevent you from travelling. However there are a few advisable steps you should take so that you make the most out of your experience.

Holiday tips:

- ✓ Make sure you stay well hydrated. It is advisable to drink purchased bottled water
- ✓ Make sure you have the recommended vaccinations. You can find recommendations for the country you are visiting at **www.fco.gov.uk** and **www.nhs.uk**. If have any concerns about vaccinations you should discuss this with your doctor
- ✓ Speak to your transplant team or nephrologist if you are planning a holiday as they can advise you on which vaccinations are safe for you to have. Remember to avoid all 'live' vaccines
- ✓ Take several days worth of medication in your hand luggage. This is just in case your luggage is misplaced by the airline because it may take several days to retrieve your luggage
- ✓ Make sure any liquid medication is clearly labelled. Current UK restrictions on taking liquids into the cabin are as follows:
 - You are allowed to take essential liquid medicines into the cabin (even if volume exceeds 100mls) but you should seek approval from the airline and departure airport.
 - You should also take a supporting letter from your doctor.
 - Further information is available at **www.direct.gov.uk/en/TravelAndTransport/Publictransport/AirtravelintheUK**
- ✓ Travel insurance should be considered as an essential part of you trip. You should always ensure you inform the insurer of your condition and any medication you take. If you do not inform them your insurance will be void. Price can vary greatly so it is advisable to compare different providers

- ✓ The European Health Insurance Card allows you to get state healthcare at a reduced cost or sometimes free of charge. Further information is available at **www.ehic.org.uk**
- ✓ More travel tips, including advice on sun care, eating and drinking abroad etc are available in the travel section at **www.nhs.uk**



My healthcare

My healthcare

'My Clinic'

When you attend your clinic appointment it is important that you make the most of your time with your consultant.

Before clinic

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.

Take your Cystagon 5-6 hours before your clinic appointment so that an accurate white cell cystine level can be taken.

If you require any assistance with communication, for example an interpreter, then contact the hospital.

During your appointment

Never be afraid to double check and ask for things to be explained if you don't understand

Ask for any words you don't 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.

Don't forget to ask the questions you have written down!

Before you leave!

Write down when your next clinic appointment is.

Ask about any support groups and if there are any events/conferences coming up.

Ask who to contact if you have any problems.

Ask how you can get hold of your blood results.

You can use the following tool to assist you:

'My Clinic'

Clinic date:

My questions:

.....
.....
.....
.....

*Any new side effects/symptoms? When did it start?
What makes it better/worse?*

.....
.....
.....

My medication (include any tablets you have bought over the counter).

.....
.....
.....
.....

My clinic summary:

.....
.....
.....
.....



Who do I contact with any problems/queries?

.....

.....

Are there any upcoming support group meetings or events?

.....

.....

How will I receive the results of my blood test?

.....

.....

The date of my next clinic appointment is:

.....

.....

Access to my records

Renal patient view (www.renalpatientview.org) allows you to access information about diagnosis, treatment and your latest blood results. Speak to your renal consultant if you are interested in signing up to renal patient view.

My care

It is important for you to have a plan of care for your condition. The following pages can be used to help plan your care over the coming months or years and provides a brief framework of what care/input you should expect to receive. You should complete your plan of care in association with a healthcare professional.

My clinic dates:

1.
2.
3.
4.
5.

My medication:

Medication name	Dose and frequency	What's it for?	Date started	Date stopped

My investigations

There are certain investigations that patients with cystinosis should have routinely to ensure they are receiving the most appropriate care. The following investigation/appointments are necessary and attendance should be discussed with your consultant.

Ultrasound scan (USS)

Date of last test

I should have this test every.....years/months*

Lung function test

Date of last test

I should have this test every.....years/months*

Echocardiogram

Date of last test

I should have this test every.....years/months*

Speech and language therapist

Date of last appointment

I should see a speech and language therapist every.....years/months*

Neurologist

Date of last appointment

I should see a neurologist every.....years/months*

Ophthalmologist

Date of last appointment

I should see an ophthalmologist every.....years/months*

*Delete as applicable

The next section is entirely up to you! If there is anything you would like to achieve, regarding cystinosis, that we can help with you can write your aims down and discuss objectives with your healthcare professional.

Aims (What you want to do)	Objectives (How it can be done)
<i>1. Example: I would like to learn more about cystinosis.</i>	<i>2. Example: Provide educational materials about cystinosis and attend conferences/events.</i>

Signature (healthcare professional)

Date

Signature (patient)

Date

