

Queen Elizabeth Hospital Birmingham



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NHS Foundation Trust



Nephropathic Cystinosis: Information for patients and families.

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www.uhb.nhs.uk/patient-information-leaflets.htm

Introduction

This information leaflet aims to help patients and carers understand cystinosis and its treatment.

What is cystinosis?

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 cells of the body.

Cystine is an amino acid. When amino acids link together they form protein which is essential to life. Protein is then broken down inside structures called lysosomes within the cells of your body.

In patients living with cystinosis, the transport system fails to carry cystine back out of the lysosomes therefore allowing 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.

- **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.

- **Adult cystinosis**

This form is sometimes described as benign cystinosis. It is usually discovered through performing other routine examinations such as an eye test. Patients with this form rarely report any symptoms with no evidence of any kidney damage.

How do you get cystinosis?

Cystinosis is a rare inherited genetic disorder. This means that it is not infectious, contagious or been brought about by lifestyle.

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.

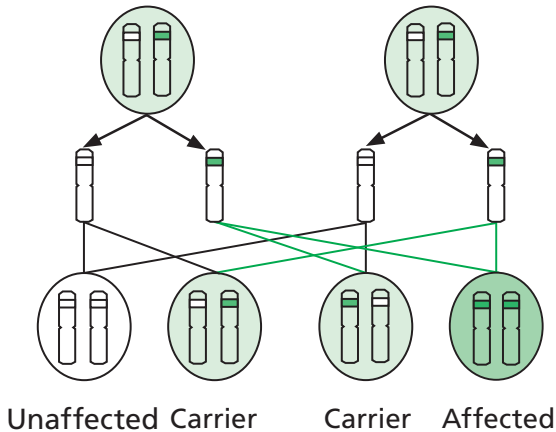
Each child of parents who both carry the cystinosis gene has a 25 % chance of inheriting a changed gene from both parents and being affected by cystinosis.

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



How is it diagnosed?

Cystinosis can be diagnosed through taking a blood test to measure the white blood cell (WBC) cystine level.

Slit-lamp examination (an eye test) can be used to check for evidence of cystine crystals in the eye. The slit-lamp is an instrument that shines a thin sheet of light into the eye.

Symptoms

In infantile cystinosis the following symptoms may occur in the first year or two after birth:

- 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 (salts and minerals that can conduct electrical impulses in the body) such as sodium, potassium, phosphorus and bicarbonate to be wasted into the urine. This is often referred to as Fanconi Syndrome.

Other symptoms, not related to the kidney, include:

- Photophobia (pain or discomfort from bright lights)
- Muscle wasting
- Accumulation of crystals in the eye
- Hypothyroidism (When the thyroid gland does not make enough thyroid hormone)
- Damage to the Pancreas, which may cause diabetes

Late onset Cystinosis displays similar symptoms which can start from the age of 2 up to the mid 20s.

Treatment

Symptomatic treatment (before kidney failure develops):

It is possible to relieve some of the 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 wasted by the kidneys.

Vitamin D and phosphorus supplements will prevent rickets.

Hypothyroidism can be treated with a hormone supplement. Insulin may be required to treat diabetes if the pancreas has been damaged.

Cystinosis Specific Treatment:

The levels of cystine within the lysosomes can be reduced by taking Cysteamine (Brand name Cystagon). Cysteamine is taken by mouth up to 4 times a day.

Cysteamine treatment reduces the progression towards renal 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.

Cysteamine 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).

Treatment monitoring

To ensure treatment is effective, it is necessary to monitor your white blood cell (WBC) cystine level. WBC levels should be checked every three to four months.

Blood samples should be taken 5-6 hours after you have taken a dose of Cysteamine. It is important to let your health care professional know if you are missing any doses of Cysteamine.

What support is available?

Cystinosis Foundation UK

Cystinosis UK is a UK based charity who aims to provide support for all those diagnosed with cystinosis, their families and their carers. They support research into the treatment of cystinosis and provide information to patients and the medical profession.

www.cystinosis.org.uk

Rare Diseases 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.

www.raredisease.org.uk

CLIMB

Climb aims to provide specific information about metabolic diseases, advice and support to children, young people, adults, families and professionals in the United Kingdom and to provide information and support to families worldwide, to fund educational and primary research programmes and to investigate treatments and medical services.

www.climb.org.uk

Genetic Alliance

Genetic Alliance UK is the national charity of patient organisations with a membership of over 140 charities supporting all those effected by genetic conditions.

www.geneticalliance.org.uk

Useful contacts

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