

Dedicated to a Cure.

Committed to Our Community.

What is Cystinosis?

Cystinosis is a metabolic disease that causes the amino acid cystine to accumulate in various organs of the body. Cystine crystals accumulate in the kidneys, eyes, liver, muscles, pancreas, brain and white blood cells.

Without specific treatment, children with cystinosis develop end stage renal failure at approximately 9 years of age. If cystinosis patients receive a kidney transplant, their new kidney will not be affected by the disease. However, without specific treatment, cystine accumulation can cause complications in other organs of the body. The complications include muscle wasting, difficulty swallowing, diabetes, blindness and hypothyroidism.

Cause of Cystinosis

Cystinosis is an autosomal recessive genetic disease. A parent of a child with cystinosis carries the gene. The parents are carriers and have no signs of the disease. The genetic mutation causes a defect in the transport of cystine out of the cells. The cystine crystallizes in the cell and destroys cells.

Our Mission

The Cystinosis Research Network is an all volunteer, non-profit organization dedicated to supporting and advocating research, providing family assistance and educating the public and medical communities about cystinosis.

Our Vision

The Cystinosis Research Network's (CRN) vision is the discovery of improved treatments and ultimately a cure for cystinosis.

Cystinosis
Research Network



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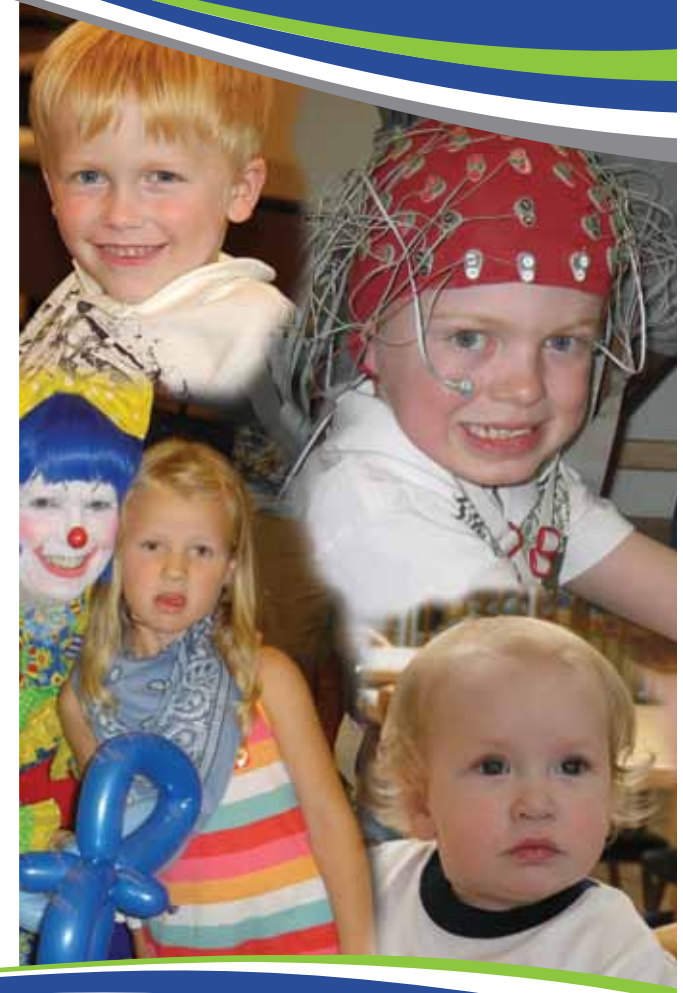
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Information presented in this brochure is intended for general education purposes only, and should not be construed as advising on diagnosis or treatment of this or any other medical conditions.



Providing Support,
Advocating Research, Family Assistance,
and Education About Cystinosis

Symptoms and Types of Cystinosis

The three types of cystinosis are **infantile**, **late onset**, and **benign**. They differ in the age of symptoms and severity.

Infantile Nephropathic Cystinosis

Symptoms:

- EXCESSIVE THIRST
- EXCESSIVE URINATION
- FAILURE TO THRIVE
- RICKETS
- EPISODES OF DEHYDRATION
- CYSTINE CRYSTALS IN CORNEA
- ELEVATED CYSTINE LEVELS IN WHITE BLOOD CELLS

These symptoms usually appear between 6 and 18 months of age. They are caused by renal tubular Fanconi Syndrome, or a failure of the kidney to reabsorb nutrients and minerals. The minerals are lost in the urine.

In **Late Onset** Cystinosis, kidney and eye symptoms typically become apparent during the teenage years or early adulthood.

In **Benign** or **Adult** Cystinosis, cystine accumulates primarily in the cornea of the eyes.



Cystinosis Treatment

Fanconi Syndrome Kidney tubular dysfunction requires a high intake of fluids and electrolytes to prevent excessive loss of water from the body (dehydration). Sodium bicarbonate, sodium citrate, and potassium citrate may be administered to maintain the normal electrolyte balance. Phosphates and vitamin D will also correct the impaired uptake of phosphate into the kidneys and to prevent rickets. Carnitine may help to replace muscular carnitine deficiency.

Cysteamine (Cystagon) is the treatment for cystinosis to reduce cystine accumulation in the cells. Cysteamine has proven effective in delaying or preventing renal failure. Cysteamine also improves growth of children with cystinosis. Cysteamine should be considered for use by post-transplant cystinosis patients.

Kidney transplantation has been an effective treatment for individuals with cystinosis who have kidney failure.

Cysteamine eye drops dissolve corneal cystine crystals and relieve photophobia. The eye drops are awaiting approval by the FDA .

Future of Cystinosis

Over the last 20 years, the prognosis of a child born with Cystinosis has greatly improved. However, now that people with cystinosis are surviving into their 20s, 30s, and 40s, new research questions need to be answered to give people with cystinosis an improved quality of life.

Scientists have mapped the Cystinosis gene, CTNS, to chromosome 17p13. Researchers are creating mouse models of cystinosis that will lead them to better understand cystinosis and develop improved treatments for each complication.

Your tax deductible contribution **will make a difference** in the fight against cystinosis.

Cystinosis Research Network, Inc.

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OR

Visit us directly online at www.cystinosis.org to make a credit card donation.

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