



CYSTINOSIS RESEARCH NETWORK

Dedicated to a Cure. Committed to Our Community.



WHAT IS CYSTINOSIS?

Cystinosis is a rare genetic, metabolic, lysosomal storage disease caused by mutations in the CTNS gene on chromosome 17p13 which results in an abnormal accumulation of the amino acid cystine in various organs and tissues of the body such as the kidneys, eyes, muscles, pancreas and brain. The cystine accumulation causes widespread tissue and organ damage. Cystine accumulation can lead to kidney failure, muscle wasting, swallowing difficulty, diabetes, hypothyroidism, cerebral atrophy, photophobia, blindness, corneal ulceration, ventilatory impairment and more. Without treatment, children with cystinosis will usually develop end stage kidney failure or die prematurely. 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.

CAUSE OF CYSTINOSIS

Cystinosis is an autosomal recessive genetic disease. A parent of a child with cystinosis carries one copy of the abnormal CTNS 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 a 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 vision is the acceleration of the discovery of a cure, development of improved treatments and enhancement of quality of life for those with cystinosis.



@CystinosisCRN



facebook.com/CystinosisResearch



@CystinosisResearchNetwork



Cystinosis Research Network



CYSTINOSIS
RESEARCH NETWORK

302 Whytegate Ct, Lake Forest, IL 60045

Toll Free 1-866-276-3669

Tel 847-735-0471 | Fax 847-235-2773

info@cystinosis.org | www.cystinosis.org

**Dedicated to the acceleration
of the discovery of a cure,
development of improved
treatments, and enhancement
of quality of life for those with
cystinosis.**



SYMPTOMS AND TYPES OF CYSTINOSIS

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

Infantile Nephropathic Cystinosis

Signs and 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. These symptoms 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.

Late Onset Cystinosis

Kidney and eye symptoms typically become apparent during the teenage years or early adulthood. Similar to nephropathic cystinosis but with delayed onset and less severity.

Ocular Cystinosis

Cystine accumulates primarily in the cornea of the eyes. No impaired kidney function or growth. Photophobia is the only symptom. Cystine crystals may be present in bone marrow as well as the cornea.



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 help to correct the impaired uptake of phosphate into the kidneys and to prevent rickets. Carnitine may help to replace muscular carnitine deficiency.

Cysteamine (Cystagon®/Procysbi®)

Cysteamine 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 also be given to cystinosis patients following kidney transplant to prevent the non-kidney complications of the disease.

Cysteamine Eyedrops (Cystaran™)

Cysteamine eyedrops dissolve corneal crystals and relieve photophobia to prevent corneal ulcerations. These eye drops are now FDA-approved as Cystaran™.

Kidney Transplantation

Kidney transplantation is an effective treatment for the kidney failure of individuals with cystinosis.

Other Treatments

Other treatments include indomethacin, recombinant human growth hormone and thyroid hormone replacement as well as treatment of bone deformities.

The psychosocial and emotional impact of the disease on patients and families is significant. Ongoing involvement with social work and mental health services may be beneficial.

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, 40s and beyond 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 and have elucidated many of the functions of cystinosis, the protein it encodes. Researchers have created mouse models of cystinosis to better understand the disease and to develop improved treatments for each complication.

