# Understanding **Cystinosis**

Leadiant Biosciences

## What is cystinosis?

Cystinosis is a rare, genetic metabolic disease.

Cystine, an amino acid, accumulates and forms crystals that can damage many organs and tissues, especially the eyes and kidneys.

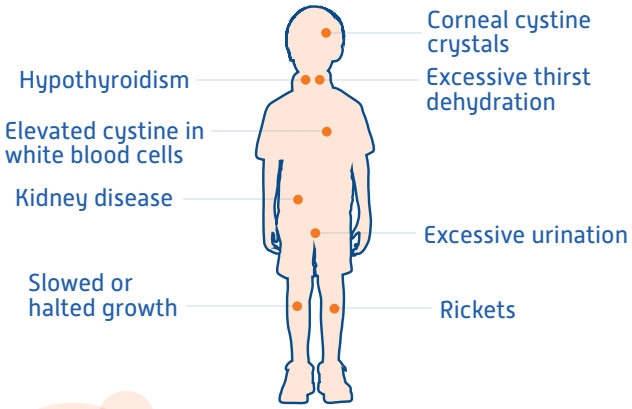
**2,000 people worldwide and 500 in the U.S. have cystinosis[1]**

## Who gets cystinosis?[1]

Cystinosis is an inherited autosomal recessive disease, meaning that a copy of the defective gene, known as CTNS, must be passed down from both parents. Parents are a carrier of the gene, but have no symptoms. Their children have a 25% chance of being affected with cystinosis.

## What are the symptoms of cystinosis?[1,2,3,4]

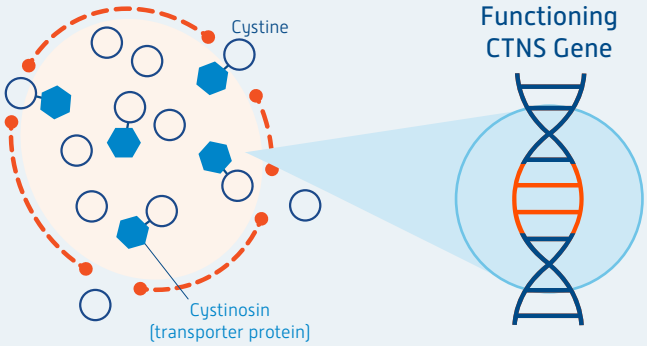
Cystinosis can affect many different parts of the body. The most common clinical findings, which begin appearing during childhood, include:




**Corneal crystals are a common clinical finding of cystinosis**

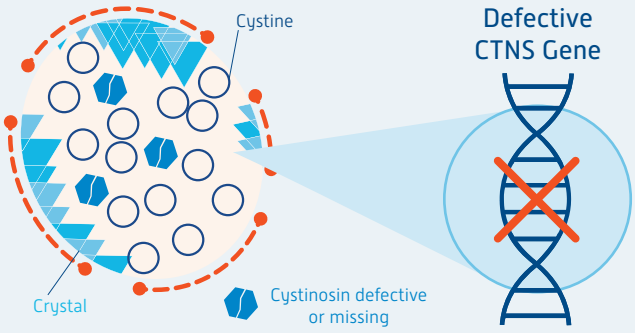
## What causes cystinosis?

Cystinosis is caused by a mutation in the CTNS gene, most commonly 57-kb deletion.




### Normal Cell

The CTNS gene provides instructions for making a transporter protein called cystinosin, which is responsible for moving the amino acid, cystine, out of the cell lysosome – the part of the cell that digests and recycles materials.[5]

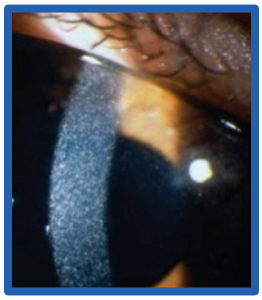



### Mutated Cell

When cystinosin is defective or missing, cystine accumulates and forms crystals in the lysosomes, damaging cells in the eyes, kidneys, and other organs.[5]

## How does cystinosis affect the eyes?[3,6,7]

Corneal crystals can be present before age one year and are always present after age 16. Left untreated, corneal crystals can lead to the following conditions and potentially cause permanent damage to the eyes:



- eye pain
- excessive sensitivity to light
- foreign body sensation in the eye
- vision haziness
- squinting
- corneal abrasion
- involuntary eye closure
- band keratophathy (a condition where a line appears across the cornea)

## How is cystinosis treated?

Treatment of cystinosis is focused on managing symptoms, as there is no cure.[1] An oral treatment called cysteamine has been shown to lower the amount of cystine in the cells, improve growth in children, and stabilize kidney and other functions.[7] However, oral cysteamine therapy does not reduce the ocular effects of cystinosis because it does not reach the corneas.[7,8]

## How are corneal crystals treated?

Corneal crystals are treated with CYSTARAN® (cysteamine ophthalmic solution) 0.44% drops. CYSTARAN is the only FDA-approved ophthalmic treatment for corneal crystals in cystinosis patients.

## INDICATION

CYSTARAN (CYSTEAMINE OPHTHALMIC SOLUTION) 0.44% STERILE is a cystine-depleting agent indicated for the treatment of corneal cystine crystal accumulation in patients with cystinosis.

## IMPORTANT SAFETY INFORMATION

To minimize contaminating the dropper tip and solution, care should be taken not to touch the eyelids or surrounding areas with the dropper tip of the bottle. Keep bottle tightly closed when not in use.
There have been reports of benign intracranial hypertension (or pseudotumor cerebri) associated with oral cysteamine treatment that has resolved with the addition of diuretic therapy. There have also been reports associated with ophthalmic use of cysteamine; however, all of these patients were on concurrent oral cysteamine.
CYSTARAN contains benzalkonium chloride, which may be absorbed by soft contact lenses. Contact lenses should be removed prior to application of solution and may be reinserted 15 minutes following its administration.
CYSTARAN is for topical ophthalmic use only.
The most frequently reported ocular adverse reactions occurring in ≥ 10% of patients were sensitivity to light, redness, and eye pain/irritation, headache and visual field defects.
Please see full prescribing information for contraindications, precautions and adverse reactions.

### References: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