

Living with Cystinosis

You or a loved one may have been diagnosed with cystinosis. This brochure provides general information about cystinosis, including how it affects the cornea of the eye.

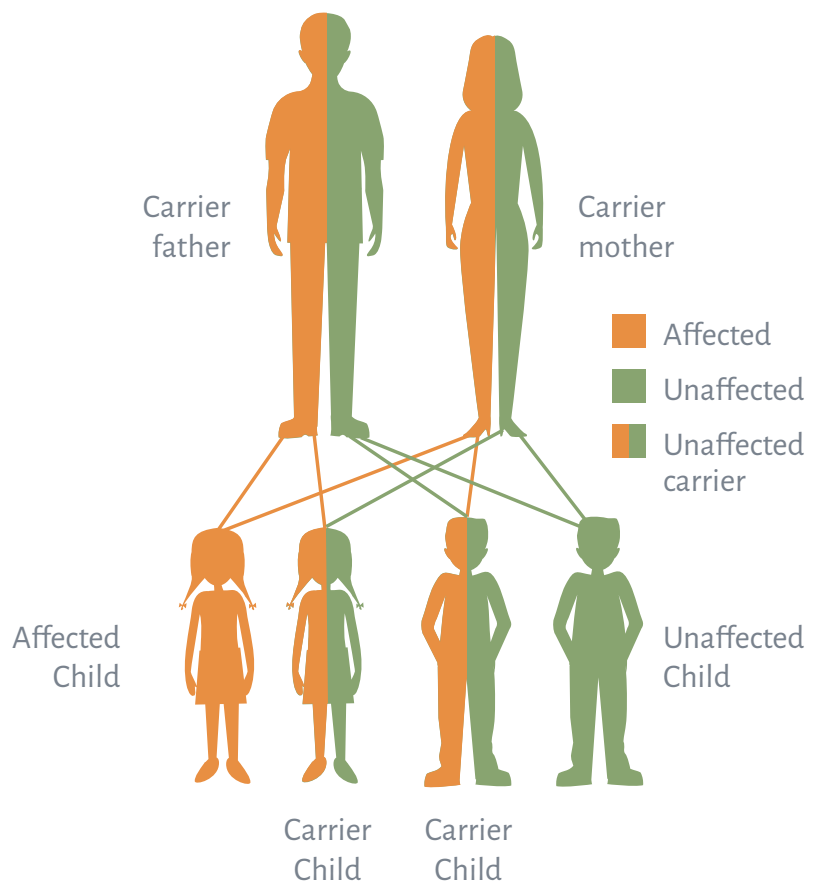


Lizzie, her parents, and her brother, Chad
(Not actual patient or caregivers)

What is cystinosis?

Cystinosis is a rare inherited disorder in which the **amino acid cystine** builds up in cells in the body and forms crystals.

More than 600 people in the United States have cystinosis. About 95% of them have **infantile nephropathic cystinosis**. (Nephropathic means diseases that involve the kidneys.) There are two other types of cystinosis – **late-onset cystinosis** and **ocular cystinosis** (non-nephropathic).



In this family, both parents are **carriers** of cystinosis because each has one unaffected *CTNS* gene and one *CTNS* gene with a mutation.

How is cystinosis inherited?

Lizzie has cystinosis because she inherited a *CTNS* gene with a mutation from each parent. The **57-kb deletion** is the most common gene variation in people with cystinosis in the United States.

When both parents are carriers, each child in the family has a 25% chance of inheriting cystinosis, a 50% chance of being a carrier, and a 25% chance of having no affected *CTNS* genes.

After Lizzie was diagnosed, Chad was tested. Like his parents, he is a carrier of cystinosis but does not have symptoms.

DID YOU KNOW? May 7th (5/7) was chosen as Cystinosis Awareness Day because of the 57-kb gene variation.

How does cystinosis affect the body?

In people with cystinosis, cystine builds up and forms cystine crystals in all cells of the body. Some organs and tissues are more likely to be impacted by cystinosis, including:

- **Kidneys**
- **Eyes**
- **Bones**
- **Muscles**
- **Thyroid** (a gland that produces hormones that your body needs)

Cystine crystals are especially damaging to the kidneys and eyes.

How does cystinosis affect the kidneys?

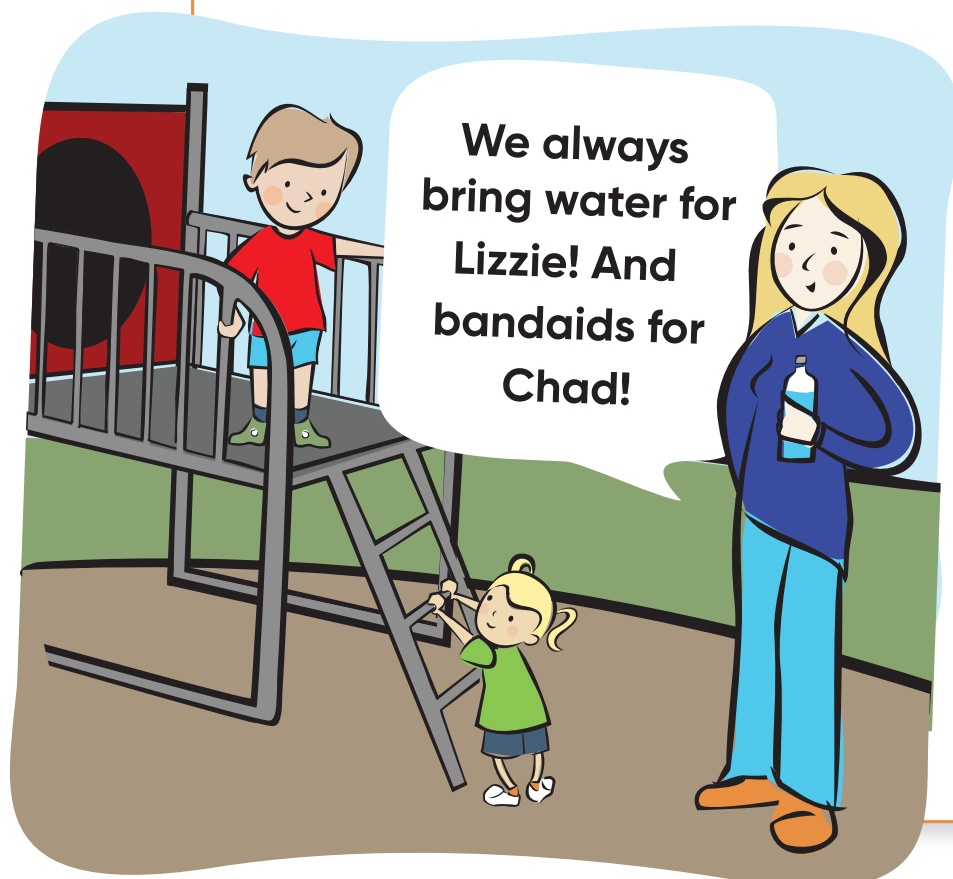
Often, the first signs of cystinosis are:

- **Urinating a lot**
- **Feeling thirsty much of the time**

These symptoms occur when the kidneys fail to reabsorb nutrients and minerals. The minerals are then lost in urine.

Frequent urination may lead to dehydration, less blood circulating throughout the body, and loss of many nutrients. Because cystinosis damages the kidneys, most people with cystinosis will eventually need a kidney transplant.

Since her diagnosis, Lizzie has been treated by a **pediatric nephrologist** – a doctor who is an expert in treating kidney diseases in children and teenagers. Lizzie's pediatric nephrologist coordinates care with other specialists as needed.



An illustration of a boy in a purple sweater and a girl in a blue shark costume. The boy is standing and looking towards the girl. The girl is also looking towards the boy. There are two speech bubbles: one from the boy and one from the girl.

Lizzie wants to be a vet when she grows up!

Shark corneas are very similar to human corneas, but sharks don't blink like humans do!

How does cystinosis affect the cornea of the eye?

In addition to seeing her pediatric nephrologist regularly, Lizzie also sees an **ophthalmologist** (a doctor who treats eye problems).

In people with cystinosis, cystine crystals may affect all parts of the eye, including the **cornea**. The cornea is the front, clear part of the eye. It is very important because it helps the eye focus, and it protects the inner parts of

the eye. The **cornea has many layers**. Over time, crystal build-up progresses deeper into the cornea, affecting all layers.

Unlike most parts of the body, the **cornea does not have blood vessels**. This means that the oral medicine Lizzie takes for her cystinosis does not reach the corneas of her eyes.

Without specific treatment for the eyes, cystine crystals will continue to build up throughout the layers of the cornea.

What eye symptoms may occur when cystine crystals build up in the cornea?

The build-up of cystine crystals in the cornea starts at birth. The crystals can be seen as early as 12 months of age by an ophthalmologist. Without treatment, symptoms caused by the build-up of cystine crystals in the cornea increase in number and intensity over time. As a result, **vision may become impaired and daily activities may become more difficult to carry out.**

Increased sensitivity to light (photophobia) is the most common eye symptom in people with cystinosis. Many people with photophobia avoid being outside, or they wear dark glasses, sometimes even inside.

Additional symptoms may develop, ranging from **squinting, uncontrollable blinking, and watery eyes to more severe symptoms** that cause discomfort and pain.



Lizzie and her parents track Lizzie's eye symptoms so that they are prepared to discuss concerns with her ophthalmologist.

Keeping track of eye symptoms

Eye Symptoms <i>Write yes or no next to each symptom.</i>	Yes/No
Light sensitivity in sunlight or other bright light	
Feeling like there is something in your eyes	
Eye pain	
Tearful eyes and/or dry eyes	
Blurry vision	
Difficulty seeing at night	
Uncontrollable blinking or winking	

Regular cystinosis medical eye exams

Regular cystinosis medical eye exams (not just vision checks) **are recommended** for people with cystinosis to assess how the amount of crystals is changing over time.

These exams are a good time to ask the ophthalmologist questions about the impact of cystinosis on the eyes, as well as to discuss other eye conditions that may have been diagnosed, such as cataracts, dry eyes, retinopathy, or glaucoma. These conditions may or may not be related to cystinosis. It's also important to let the ophthalmologist know about any extended periods of time that oral or eye medicines were not used.

For better coordination of care, **it's important for all doctors to receive up-to-date results of eye exams** to help ensure the best possible treatment plan moving forward.

Questions to ask your ophthalmologist

- ✓ **How is cystinosis affecting my eyes?**
- ✓ **What parts of my eyes are affected the most?**
- ✓ **What is the amount or density of cystine crystals in my corneas?**
- ✓ **Has there been a change in the amount of cystine crystals since my last exam?**
- ✓ **How would you rate my photophobia?**
- ✓ **Are my eye symptoms due to the effects of cystinosis on my corneas or on other parts of my eyes?**
- ✓ **What can I do to prevent long-term complications of cystinosis in my eyes?**

DID YOU KNOW? Your ophthalmologist can take pictures of your corneas to assess the build-up of corneal crystals over time.

A personalized treatment plan for a systemic disorder

Lizzie has a personalized treatment plan that she follows every day to help her stay healthy. This includes:

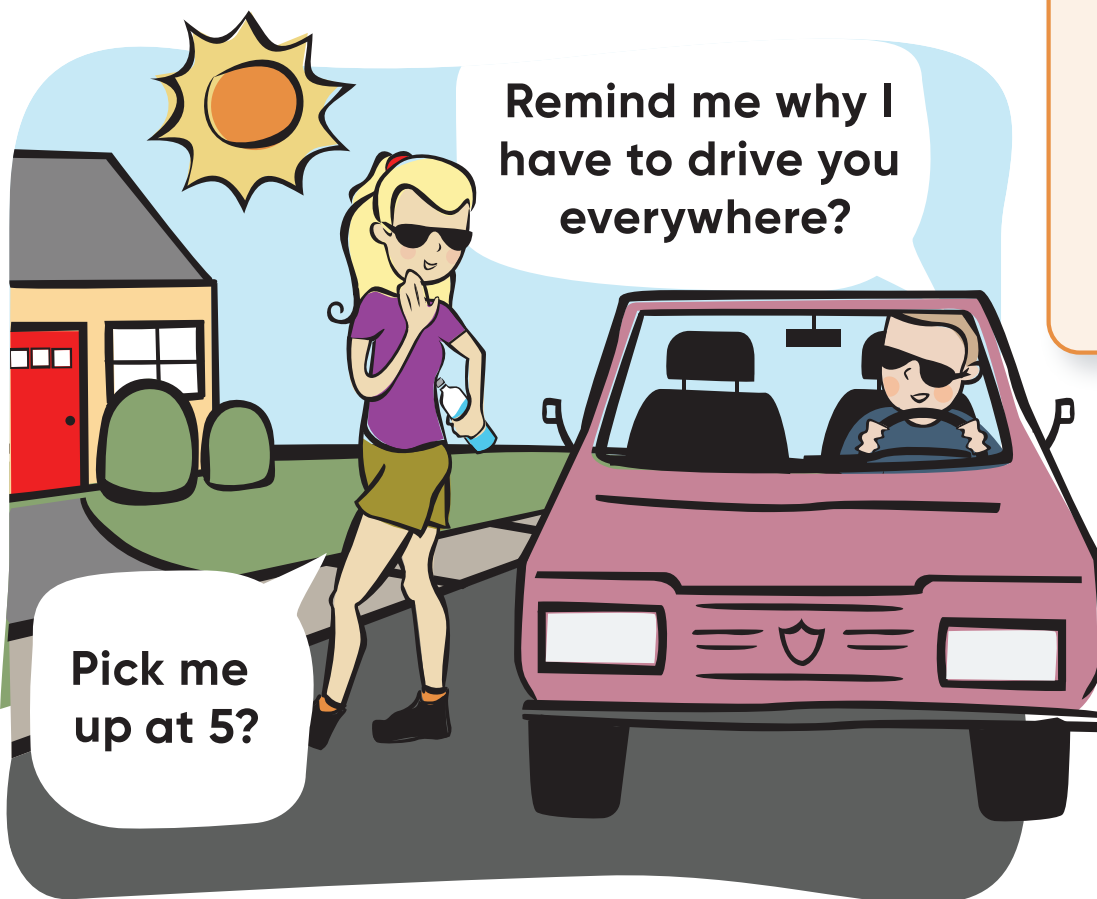
- **Taking medicines that reduce the build-up of cystine crystals in the body, including the eyes**
- **Taking medicines and nutritional supplements that prevent or treat symptoms caused by cystine crystal build-up**

At every appointment, Lizzie talks to her doctors about things that may get in the way of keeping up with her medication schedule, such as school, after-school activities, work, and vacations. Importantly, they also talk about how Lizzie can manage her cystinosis while still doing the things she wants to do.

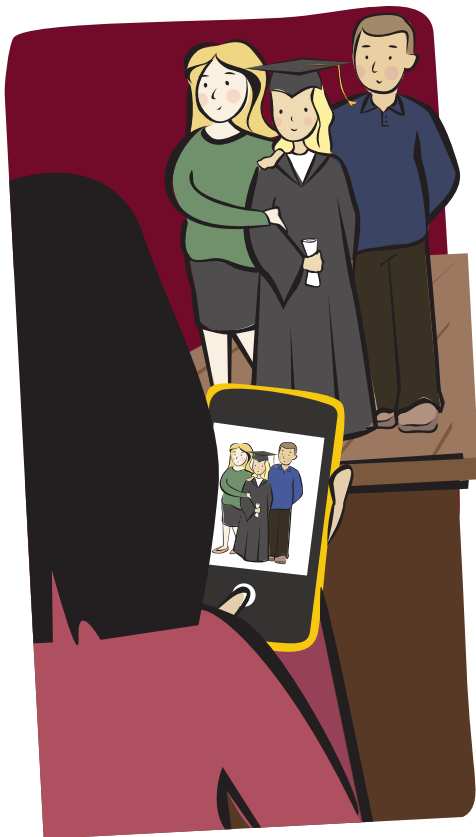
Lizzie also has a water bottle with her all the time because she needs to drink a lot, and she makes sure to exercise and eat a healthy diet.

DID YOU KNOW?

Cystinosis is among the approximately 3% of rare genetic disorders for which there are specific medicines approved by the Food and Drug Administration (FDA).



Lizzie takes her medicines on a schedule, but it's her social schedule that keeps *everyone* busy.



A new chapter

As Lizzie gets older, she will transition to seeing a **primary care doctor and other specialists for adults**.

It's important to find a doctor who acts as a partner and advocate. The Cystinosis Research Network maintains a list of **nephrologists and other healthcare providers who have training and experience in treating people with cystinosis**. Go to **Doctors in Your Area (cystinosis.org)** where you can search by provider type and geographic location.

As a young adult, Lizzie will be learning more about cystinosis and how to manage it by talking to her current care team before transitioning to adult care. Over time she will start making her own healthcare decisions.

Being able to answer the questions below can help Lizzie prepare to transition to a new medical team.

- ✓ **What is cystinosis?**
- ✓ **When were you diagnosed with cystinosis?**
- ✓ **What are your major symptoms and complications due to cystinosis?**
- ✓ **What medicines are you taking, and how frequently are you taking them?**
- ✓ **What specialists do you see, and how often do you see them?**
- ✓ **What kinds of tests do you need to track changes in cystine crystals in your body and in your eyes?**
- ✓ **Do the specialists on your treatment team belong to the same healthcare system?**
- ✓ **Do the specialists on your treatment team communicate with each other? If not, how can you facilitate communication between your doctors?**

As someone with cystinosis, Lizzie will need to schedule doctor and ophthalmologist appointments to **monitor the cystine levels in her blood and the cystine crystals in her corneas**. Staying on top of her disease will help Lizzie to be independent without compromising her health.

Staying connected

Staying connected and having social support may help to improve well-being and the ability to manage challenges.

Many people with chronic diseases benefit from being part of a **support group**. The **Center for Chronic Illness** (www.thecenterforchronicillness.org) sponsors a web-based support group for adults with cystinosis called **Living with Cystinosis**.



Looking ahead

Many people with cystinosis lead full, productive lives. Lizzie loves being a vet, and her furry patients love her back!

DID YOU KNOW? There are helpful resources available for people with cystinosis and their families. Check out the **Cystinosis Research Network (CRN)** at: www.cystinosis.org.

