

What is cystinosis?

Whether your cystinosis journey has just begun or you've been living with the disease for a long time, understanding what's going on in the body can help. Cystinosis is a rare, genetic disease. It's caused by a buildup of cystine. This buildup causes crystals to form and harm the body.

Cystinosis is...

RARE.

About 500 to 600 people in the United States have cystinosis. An estimated 20 new cases of cystinosis are diagnosed each year.



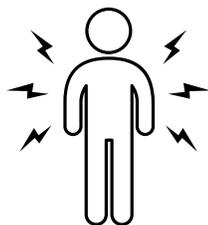
GENETIC.

Cystinosis is inherited, which means it's passed down in a person's genes. Some people may be carriers of the disease. Carriers don't have the disease, but they can pass it on to their children.



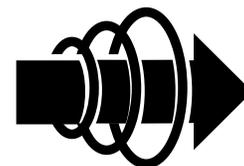
HARMFUL TO THE WHOLE BODY.

In people with cystinosis, an amino acid called cystine gets trapped inside cells. This causes harm or damage to every cell and organ in the body. A person may not feel the damage, but it can be happening even if there are no signs or symptoms.



PROGRESSIVE.

Cystinosis gets worse over time, and damage to cells cannot be undone. The goal of treatment is to slow down damage to the body by reducing cystine buildup in cells.



The 3 types of cystinosis

Nephropathic or **classic infantile cystinosis** is the most common form of the disease. It is also the most serious. About 95% of people with cystinosis have this type. Symptoms usually appear early in the first year of life, and it's a lifelong disease.

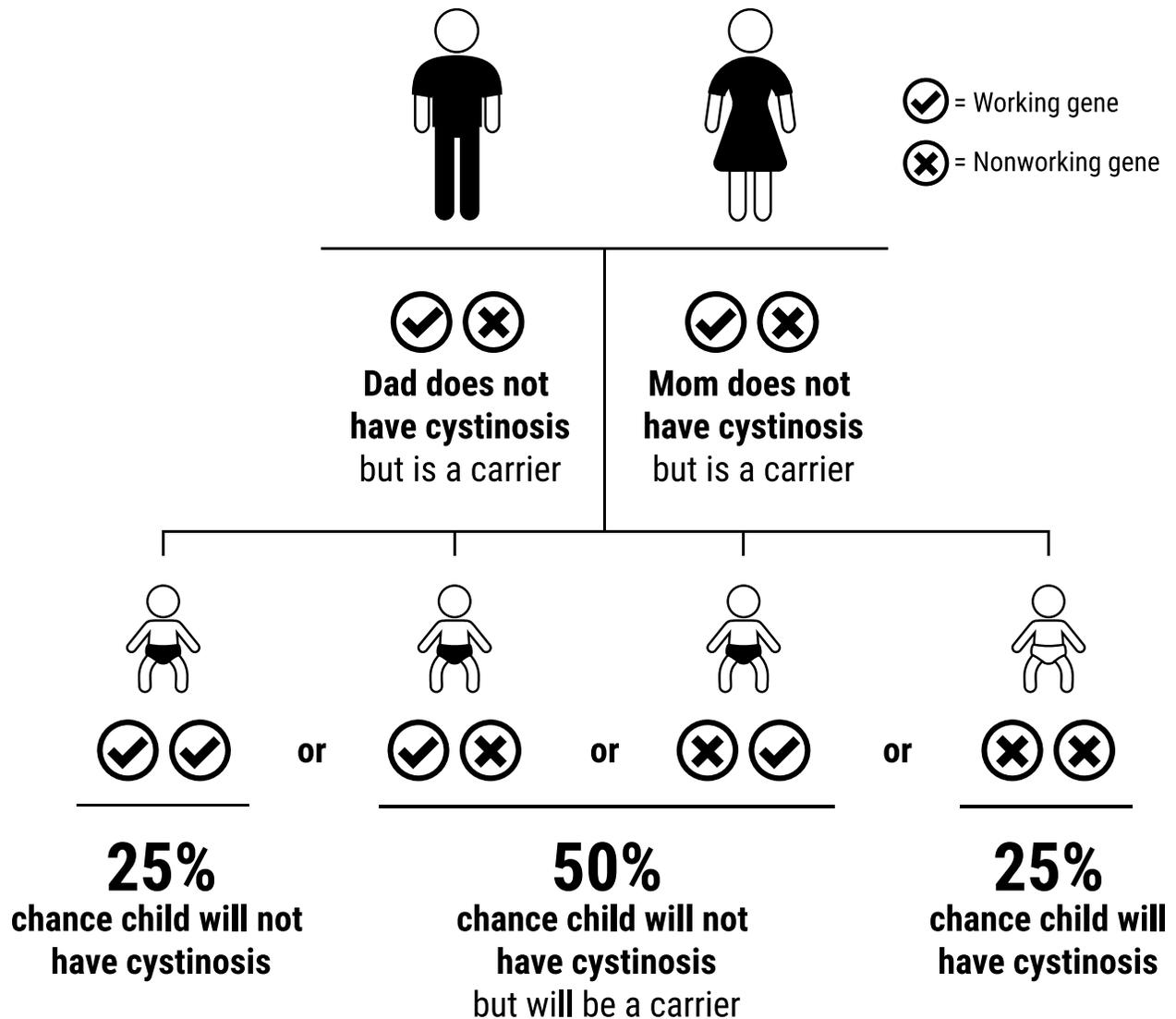
Intermediate or **juvenile cystinosis** is a less serious form of the disease. It may not be diagnosed until a person is a teenager.

Ocular or **nonnephropathic cystinosis** is the least serious form of the disease and only affects the eyes.

How cystinosis is passed down

Cystinosis isn't something you can catch from another person. It's a genetic condition, which means a child is born with it. A child can get cystinosis if both parents are carriers of the disease. A child gets cystinosis when the specific gene that doesn't work right is passed down from both parents.

Chances of a carrier couple having a child with cystinosis



When two carriers have a child, there is a 25% chance that the child will have cystinosis and a 50% chance of the child being a carrier.

How cystinosis affects the body

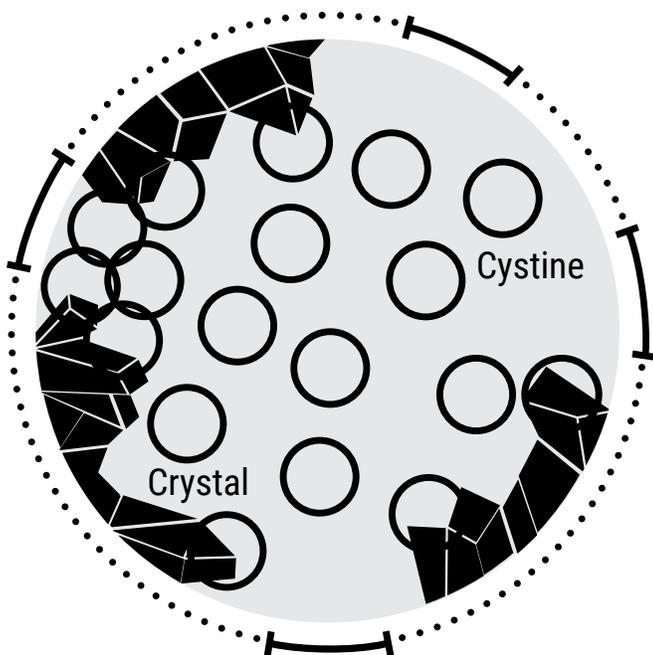
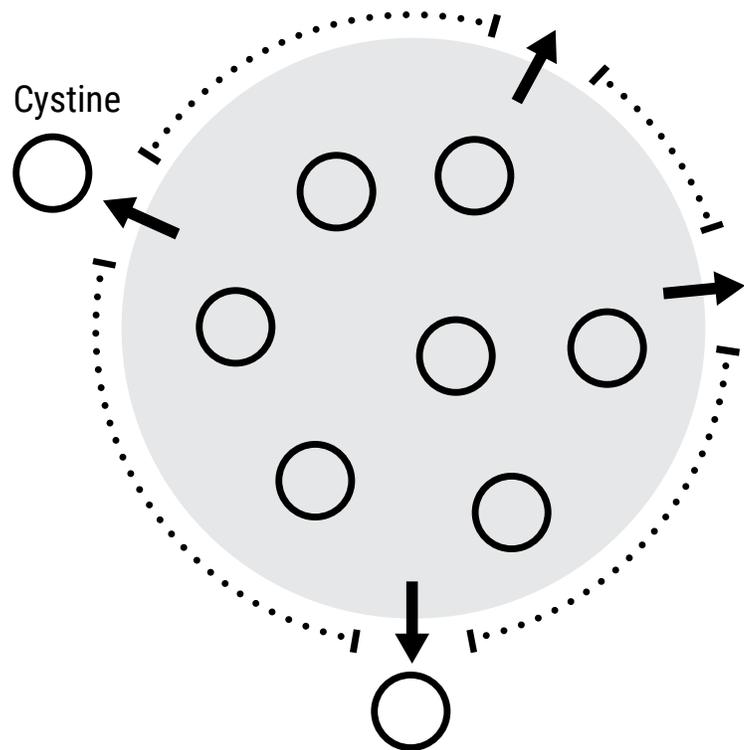
In people with cystinosis, cystine gets trapped in the cells. This causes cystine levels to rise. When cystine levels rise, crystals form that lead to harm to the body.

What cystinosis looks like in the cells

Human cells are like little factories in the body. They do a lot of work to help keep the body healthy. But in people with cystinosis, a part of the cell called the “lysosome” doesn’t work right.

Lysosome in a person without cystinosis

In a person without cystinosis, cystine is removed from the lysosome and used normally.



Lysosome in a person with cystinosis

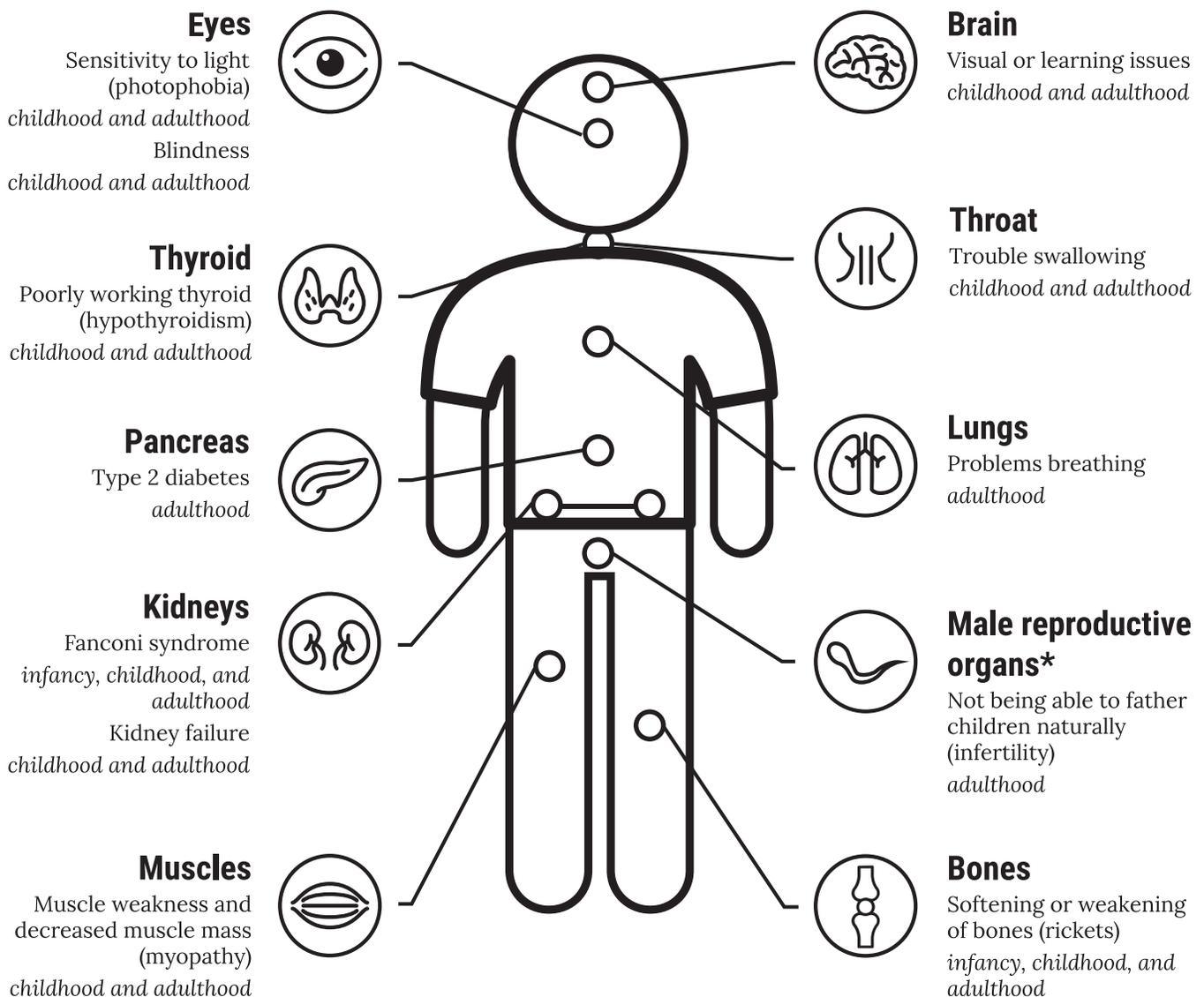
In a person with cystinosis, cystine can't be removed, so it builds up and forms crystals. These crystals lead to damage in cells and organs all over the body.

How cystinosis affects the body (continued)

How cystinosis can harm the body over time

Cystinosis harms the body little by little over time. This means that new cystinosis issues can appear at different ages.

Cystinosis can also progress differently in each person. The age when new damage appears can be different for every person. The amount of damage to the body and the rate at which it occurs is related to how well a person manages his or her condition.



*Women of childbearing age with cystinosis may become pregnant. If you are pregnant, planning on becoming pregnant, and/or breastfeeding, talk with your doctor about which treatments may be right for you.

How cystinosis affects the body (continued)

Fanconi syndrome and cystinosis

Fanconi syndrome is a type of kidney disease. It is often the first sign of cystinosis. It can lead to:

- The body losing important substances needed for good health
- Feeling the need to pee (urinate) a lot
- Extreme thirst or dehydration
- A softening or weakening of the bones, commonly called rickets

For more information about our community of people living with cystinosis, visit CystinosisUnited.com. You'll also learn more about what causes this rare genetic disease, signs and symptoms, ways to manage, treatment options, and the latest tips and advice.