

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 may help. Cystinosis is a rare, genetic disease. It's caused by a buildup of cystine in the body. This buildup causes crystals to form and harm the body.

Understanding cystinosis

Cystinosis is...

RARE

Approximately 100 people in Canada have cystinosis.



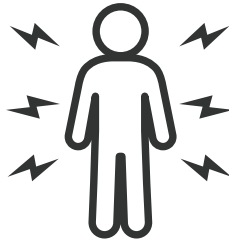
GENETIC

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 may happen without 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 cystinosis 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.

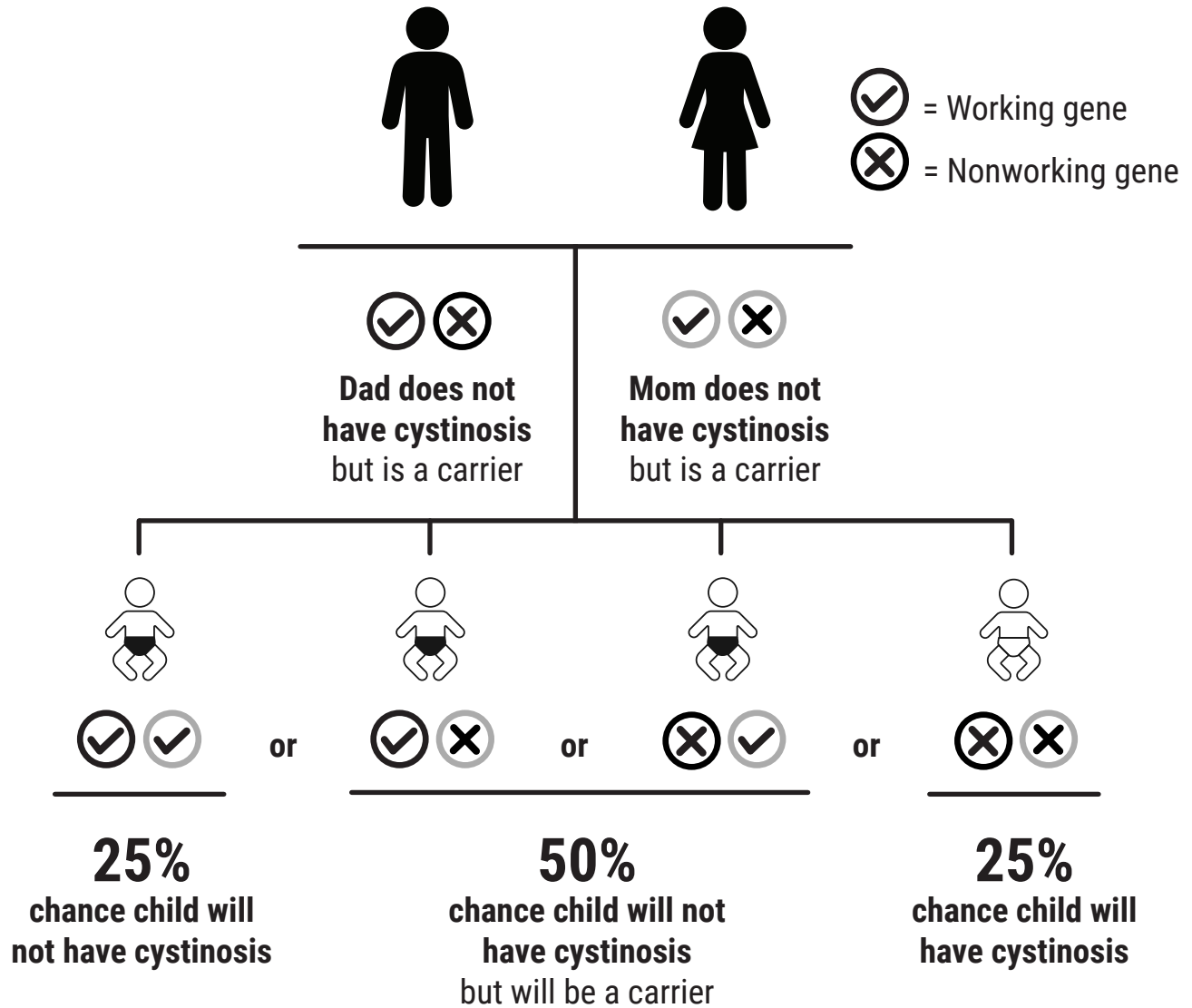
Intermediate cystinosis or **juvenile cystinosis** is a less serious form of the disease. It may not be diagnosed until a person is a teenager.

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

How people inherit cystinosis

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 gets cystinosis if both parents are carriers of the disease and the gene that doesn't work right gets 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

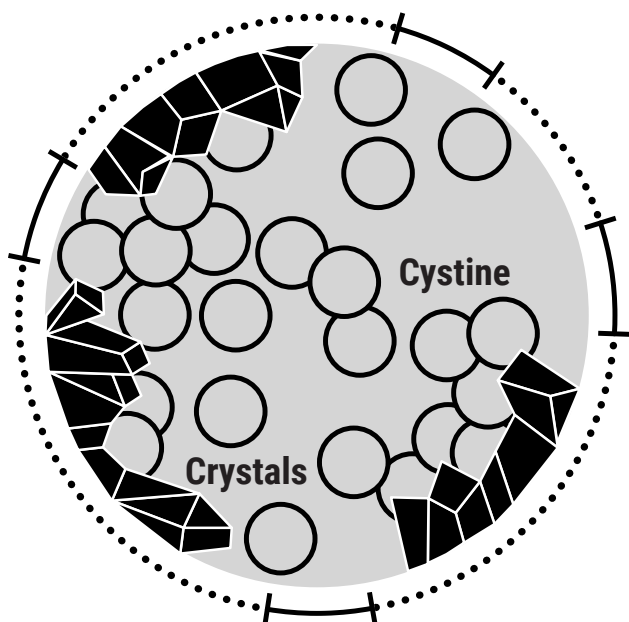
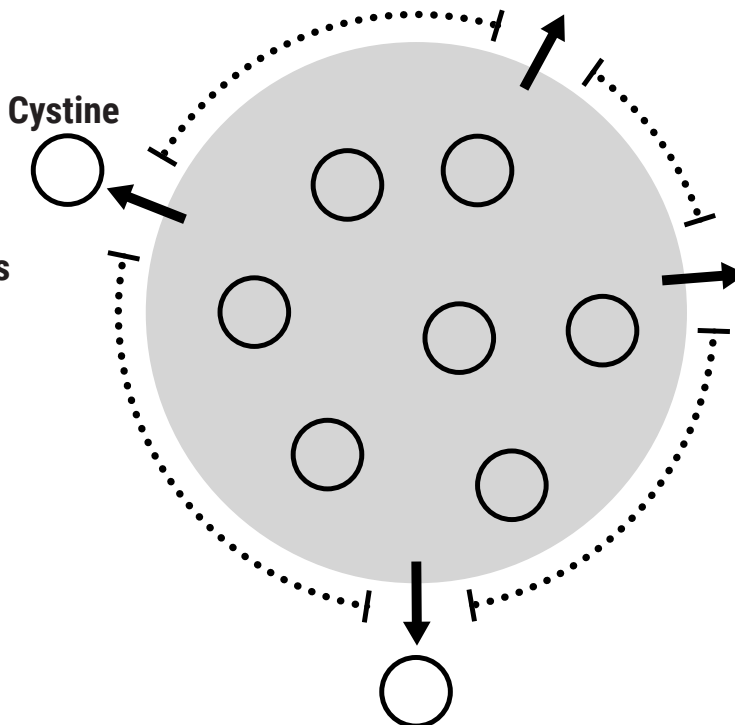
In people with cystinosis, cystine gets trapped in the cells. This causes cystine levels to rise. When cystine levels rise, crystals form that lead to harm to the body.

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 cells called the “lysosome” doesn’t work right.

Lysosome in a person without cystinosis

Cystine is removed from the lysosome and used normally.



Lysosome 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.