What is Cystinosis?
Cystinosis is an inherited disease characterized by a type of kidney disease called renal tubular Fanconi syndrome, poor growth, and photophobia (sensitivity to light in the eyes).\(^1\) It involves abnormalities in the protein cystinosin, which moves the amino acid cystine within cells. Symptoms associated with cystinosis are due to a build-up of cystine, which forms crystals within cells throughout the body, resulting in tissue damage. Cystinosis is also known as cystine storage disease.\(^2\)

What are the symptoms of Cystinosis and what treatment is available?
Cystinosis is a disease that varies in severity and age of diagnosis. There are three types of cystinosis, characterized by age of onset and kidney involvement. The most severe type is nephropathic cystinosis that begins during infancy, with symptoms that may include\(^1\):

- Poor growth, often noted by six to nine months
- Renal tubular Fanconi syndrome, as early as six months; can lead to kidney failure by age 10 if untreated
- Rickets (softening of the bones)
- Myopathy (muscle wasting and weakness)
- Acidosis (abnormally acidic blood)
- Photophobia (light sensitivity) that may lead to blindness if untreated
- Diabetes, thyroid, and nervous system problems

Intermediate cystinosis includes the same symptoms as the nephropathic type, but it begins later in childhood or adolescence. If untreated, kidney failure will result by age 15 to 25. Individuals with nonnephropathic cystinosis typically have normal growth and kidney function, and the signs of disease are limited to the eyes.\(^1\)

There is no cure for cystinosis and, if left untreated, nephropathic and intermediate cystinosis will result in an early death. Medications to reduce the accumulation of cystine crystals throughout the body are available. Medication use may decrease the kidney and growth problems associated with this disorder but may not reverse the damage. Kidney transplantation may be considered for some individuals. Additional treatment may include increased water intake, nutritional supplements, growth hormone therapy, and speech and physical therapy.\(^1\)

How is Cystinosis inherited?
Cystinosis is an autosomal recessive disease caused by mutations in the \textit{CTNS} gene.\(^2\) An individual who inherits one copy of a \textit{CTNS} gene mutation is a “carrier” and is not expected to have related health problems. An individual who inherits two mutations in this gene, one from each parent, is expected to be affected with cystinosis.

If both members of a couple are carriers of a mutation in the \textit{CTNS} gene, the risk for an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

Who is at risk for Cystinosis?
Cystinosis can occur in individuals of all races and ethnicities, but it appears to be most common in individuals of French Canadian ancestry, who have an estimated carrier frequency of 1 in 39.\(^3\) Worldwide, cystinosis is estimated to affect 1 in 100,000 individuals with a calculated carrier frequency of 1 in 158.\(^2\)
Having a relative who is a carrier or who is affected can increase an individual's risk of being a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

**What does a positive test result mean?**
If a gene mutation is identified, an individual should speak to a physician or genetics health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

**What does a negative test result mean?**
A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

**Where can I get more information?**

**References**