CYSTINOSIS

Cystinosis is a rare genetic disease that causes an amino acid called cystine to build up inside your cells. This leads to the formation of crystals that can damage the body's organs and tissues. In time the kidneys, thyroid, pancreas, muscles, eyes, and many other parts of your body can be affected. The more cystine collects in your cells, the more severe the disease. There are three forms:

- ***Infantile (early-onset) cystinosis*** is by far the most common and most severe. Symptoms begin to appear between 6 and 12 months of age, and may include feeding problems, below-average growth, constant thirst, and frequent urination. Eventually the kidneys become less able to absorb nutrients and minerals, and if it's not properly treated this form of cystinosis usually leads to kidney failure by the age of 12. Other problems develop over the course of the disease as well, including difficulty swallowing, muscle wasting, weaker bones, and increased risk of diabetes and hypothyroidism. If cystinosis affects the brain, children can have problems with coordination, memory, and attention.
- In ***adolescent (late-onset) cystinosis,*** the buildup of cystine in the body's tissues is slower and symptoms develop later. If the disease is left untreated, kidney failure can develop by the late teens or early 20s.
- ***Adult cystinosis*** is less severe and is associated mainly with a buildup of cystine in the eyes, which causes discomfort and a sensitivity to light. Adult cystinosis does not usually lead to kidney failure.

What causes cystinosis?

Cystinosis is an inherited disease. When an abnormal copy of the CTNS gene is passed on from both parents, the child will have cystinosis. The form and severity of the disease will depend on the types of mutations in each parent's CTNS gene.

How is cystinosis diagnosed?

There are two tests that will confirm that a person has cystinosis. The first is a blood test to measure the amount of cystine in a specific type of white blood cell. The second is a genetic test that can identify abnormal versions of the CTNS gene.

Doctors will also perform a physical exam to look for signs of the disease, such as cystine crystals in the eyes. Blood and urine tests may be ordered to check how well the kidneys and other organs are working.

How is cystinosis treated?

Cysteamine therapy treats the disease directly by reducing the amount of cystine in the cells. If it's started as soon as diagnosis is confirmed, it can delay kidney failure, hypothyroidism, diabetes, and other problems caused by the disease.

There are also treatments that focus on improving the symptoms that develop in the parts of the body affected by cystinosis. These treatments may include:

- Supplements to replace electrolyte and other nutrient losses.
- Hormone replacement therapy for children with hypothyroidism.
- Growth hormone therapy.
- Insulin and other medications to treat diabetes.
- Eye drops to treat and prevent the formation of crystals on the cornea.
- A kidney transplant or dialysis for children with end stage renal disease.

Cystinosis is a very serious disease with a lifelong impact, but people with cystinosis today live longer into adulthood than ever before when they are treated early and consistently.

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