Cystinosis 101

Cystinosis is a rare genetic, metabolic, lysosomal storage disease caused by mutations in the *CTNS* gene on chromosome 17p13 which results in an abnormal accumulation of the amino acid cystine in various organs and tissues of the body such as the kidneys, eyes, muscles, pancreas and brain. The cystine accumulation causes widespread tissue and organ damage. Cystine accumulation can lead to kidney failure, muscle wasting, swallowing difficulty, diabetes, hypothyroidism, cerebral atrophy, photophobia, blindness, corneal ulceration, ventilatory impairment and more. Without treatment, children with cystinosis will usually develop end stage kidney failure or die prematurely. If cystinosis patients receive a kidney transplant, their new kidney will not be affected by the disease. However, without specific treatment, cystine accumulation can cause complications in other organs of the body.

Learn More About

Symptoms & Types of Cystinosis

Diagnosis

Inheritance/Genetics

Symptoms and Types of Cystinosis

There are three types of cystinosis that differ in the age of onset and severity of symptoms. They are:

1. Infantile Nephropathic
2. Late Onset
3. Ocular

Infantile Nephropathic Cystinosis

Symptoms usually appear between 6 and 18 months. Symptoms of renal Fanconi syndrome are evident at that time.

Signs and Symptoms:

Excessive thirst

Excessive urination

Failure to thrive

Rickets

Episodes of dehydration

Cystine crystals in cornea

Elevated cystine levels in white blood cells

These symptoms are caused by renal tubular Fanconi syndrome, or a failure of the kidney to reabsorb nutrients and minerals. The minerals are lost in the urine.

In **Late Onset Cystinosis** kidney and eye symptoms typically become apparent during the teenage years or early adulthood. Similar to infantile nephropathic cystinosis but with delayed onset and less severity.

In **Ocular Cystinosis** cystine accumulates primarily in the cornea of the eyes. No impaired kidney function or growth. Photophobia is the only symptom. Cystine crystals may be present in bone marrow as well as the cornea.

Diagnosis
Children usually display symptoms by 9 months of age and can be diagnosed. Unfortunately, many children are misdiagnosed and suffer for years or die before an appropriate diagnosis is made.

Diagnosis can be made by:
Fanconi syndrome symptoms and documentation of urinary losses of glucose, essential electrolytes and minerals, and amino acids

Blood test measuring elevated white cell cystine levels

Eye exam with slit lamp to confirm crystals on the surface of the eye (cornea)

Learn about treatment and how to manage cystinosis.

Inheritance/Genetics
Every person's body is made up of millions of tiny structures called cells. Each cell comes with a full set of instructions which tell the cell what to do and how to make our bodies work. The instructions are called genes, and they are made from a chemical called DNA. Genes usually come in pairs, and they determine everything about our bodies. For example, certain genes determine the color of our eyes, while other genes determine our blood type.

Genes are often called the units of heredity because the information they contain is passed from one generation to the next. We all get one gene in each pair from our mothers and the other gene in the pair from our fathers. In this way our bodies work with a combination of instructions inherited from both our parents. Parents have no control over which genes get passed to their children.

Cystinosis is an autosomal recessive genetic disease. A parent of a child with cystinosis carries one copy of the abnormal *CTNS* gene. The parents are carriers and have no signs of the disease. The genetic mutation causes a defect in the transport of cystine out of the cells. The cystine crystallizes in the cell and destroys cells.

Each time two such cystinosis carriers have a child together, there is a 1-in-4 chance (25% risk) of having a child with cystinosis. and every healthy sibling of a child with cystinosis has a 2-in-3 chance (66% risk) of being a carrier, like his parents.