



Patient Information

Cystinuria

This leaflet provides information about cystinuria.

What is cystinuria?

Cystinuria is an inherited disorder in which the urine contains abnormally large amounts of cystine. Cystine is an amino acid. There are twenty different amino acids. Amino acids are the building blocks of proteins.

Normally large amounts of amino acids pass from the blood into the kidney and then back into the blood again.

In cystinuria the transporter which transports cystine (along with ornithine, arginine and lysine) into the blood is faulty. Therefore large amounts of cystine, ornithine, arginine and lysine are lost in the urine.

Why does it matter?

Cystine does not dissolve well. If the concentration in urine becomes very high, crystals form and clump together. Eventually these make kidney stones.

The only risk with cystinuria is kidney stone formation. Although the condition is present from birth, stones generally become evident between 10 and 30 years of age. The risk is present throughout life. This disorder does not cause any other problems - affected people are otherwise quite normal.

How is it diagnosed?

This may be from chemical analysis of a kidney stone. The diagnosis is confirmed by analysing the amino acids in urine and finding large amounts of cystine, ornithine, arginine and lysine.

How is it treated?

The aim is to keep cystine dissolved in urine. The key treatment is to drink 6 to 8 pints of liquid, evenly spaced throughout the 24 hours (i.e. at least 1 pint every 4 hours). It is essential to have a drink during the night. The urine must never become concentrated: it should look like water, and never be yellow.

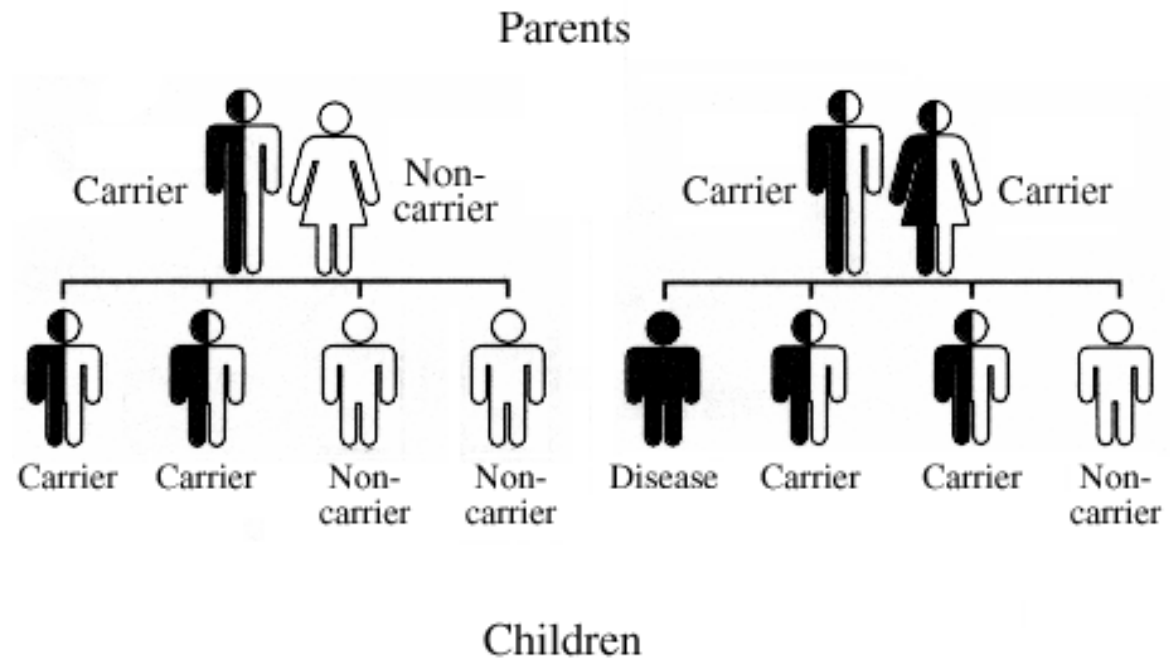
Often it is necessary to take an alkali such as potassium citrate. Cystine dissolves less well when the urine is acid. We measure acidity as pH - a low pH value (6 or less) indicates an acid urine whereas a pH value greater than 7 indicates an alkaline urine. Cystine is most soluble when the pH is about 7.5.

Together, these two treatments should stop new stones forming. Sometimes, unfortunately, stones continue to form. In this situation there is another treatment available. This involves taking D-penicillamine tablets. This substance links up with cystine in urine and helps it to dissolve. D-penicillamine can cause side effects (see separate information sheet), which generally disappear when treatment is stopped.

How is it inherited?

Cystinuria is an autosomal recessive disorder. Inherited diseases are determined by two genes. In a recessive disorder both genes are abnormal. People who have one abnormal gene and one normal gene are called carriers. They do not have the disorder. Both the parents of a person with cystinuria are usually carriers. Children get one gene from their mother and one gene from their father.

Each child of two carriers has a 25% chance of inheriting the disorder (both genes abnormal), a 25% chance of having two normal genes, or a 50% chance of being a carrier (one normal and one abnormal gene).



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