Cystinosis, the Early Years: Fanconi Syndrome and More

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The family of a child with cystinosis must deal with many challenges in the first decade of life. Fanconi syndrome leads to substantial losses of water and important electrolytes in the urine. This requires high intake of fluid and multiple medications to replace these losses. Dehydration is a frequent complication in children with cystinosis. Weight gain and growth are usually slow. This often requires a variety of strategies to increase intake of calories and some children benefit from receiving growth hormone injections to improve growth. Children usually have evidence of rickets (weak bones) at the time of diagnosis, and some have deformities of their legs that may require the input of an orthopedist.

The other challenge is the need to take a medication called cysteamine to treat the cystinosis. While cysteamine slows the deterioration in kidney function and prevents multiple other complications of cystinosis, it is an extremely challenging medication since it often causes nausea and vomiting. Cysteamine also causes many children to have an unpleasant smell.

Most children require a gastrostomy-tube for medications and supplemental fluid and calories early in life, though it frequently can be removed when the child is older and can take pills. Children also need to use cysteamine eye drops to dissolve crystals in the eyes that cause excessive sensitivity to light ("photophobia").

In general, care of children with cystinosis gets easier as they get healthier and older. New challenges may include dialysis and transplantation, though these are typically not needed until after the first decade of life.