

Presentation Day 1

Cystinosis Medication Management Throughout the Life Course

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When an infant is newly diagnosed with cystinosis, the incredibly complicated regimen, numerous medications, fluid, nutrition, and frequent dosing are overwhelming. That time and mental effort this takes affects the whole family and extended family. There are some tips and tricks that can be useful to help this go a little smoother. Being part of the cystinosis community on social media and developing a mentoring relationship with another, more experienced family can be really helpful. Some knowledge which has been gained by trial and error isn't immediately handed out to new families, because their doctors, in spite of their best intentions, may not be experts in cystinosis. Some of these suggestions will be useful to some families but not to others.

Late childhood and middle school are hopefully periods of quiet stability, as the family routine has become a well-oiled machine. As children enter the teen years, the previous well-oiled routine may fall apart as children develop a new sense of self and autonomy as young adults. This can be terribly distressing for the family because nonadherence with kidney transplant medications may have critical and irreversible consequences, yet you have to give your emerging adult enough rope to make some mistakes (but with a very tight safety net).

Adults living with cystinosis may have separate problems. Many years of chronic kidney disease or returning to dialysis after a failed kidney transplant or diabetes all may cause difficulties with stomach emptying and that can lead to complications with medication taking, increased gastrointestinal symptoms and being unable to take a full dose of cystine depleting therapy amongst other medications. Timing of medications may be important, investigation for delayed gastric emptying or dysmotility may be very useful. In some patients, placing a tube directly into the jejunum allows nutrition and medications to be taken with much less abdominal pain and improve quality of life.