The Changing Face of Cystinosis From Childhood to Adulthood

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After the initial diagnosis of cystinosis and the first few years of treatment, children face adolescence, the teenage years and finally adulthood. Many things change at an individualized pace. Liquid medications give way to pills and if a gastrostomy tube is used, they may be used less as the older child takes medications, food, and fluids orally and relies less on the G-tube. Parents learn how their child can change as they progress from the early years (including the frustrating fours) towards the independence of the (terrifying) teenage years. Then adulthood "happens" with too many seemingly abrupt changes (education, medication insurance, social supports, adult health care) that appear to be intent on disrupting everything!

Treatment of the issues faced by those living with cystinosis is complex given the changing face of the disease over time. Care plans often require a staged approached, working with families and individuals to both prioritize and focus on different goals at different times in their journey. With the initial presentation of cystinosis, there is a strong focus on weight gain, growth, cysteamine and supplements as patients often present with what seems like unending nutrition and electrolyte difficulties. As these issues settle and growth stabilizes, lower kidney function and proteinuria may need attention. Cystinosis can affect other organs and systems that can give rise to other issues, hopefully only in adulthood, if at all. Continued monitoring of corneal crystals, thyroid function, muscle strength, bone density, vitamin D and parathyroid hormone levels, glucose control, gonadotrophins, copper levels, iron levels and other parameters may give reassurance that these organs and systems are unaffected or not requiring additional intervention. Fertility and pregnancy have challenges that should be addressed. As with any chronic disease, mood and self-esteem may be an issue at any age.

Kidney transplantation seems to be inevitable despite the very best WBC cystine level control. Managing the unique issues of a low GFR in someone with cystinosis requires an understanding of the continued role of potassium and phosphate supplementation (almost never required with severe chronic kidney disease) and the beneficial effects of post-transplant native nephrectomies given a high urine output. Transplant centres will be very familiar of the usual post-transplant issues, e.g. hypertension and cardiovascular risk factors, but generally not in the other medical issues that people with cystinosis may have.

In the same way that parents are their child's experts and should always be listened to, adults living with cystinosis may well be the local expert and be a valued partner in managing the adult aspects of cystinosis. This may require multiple clinics but hopefully there will be a medical "home" that can quarterback the continuing journey.

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