Living well with cystinosis after kidney transplantation

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Living well with cystinosis after transplantation depends on four interacting factors. First preparation for transplantation and gaining understanding/knowledge about the medical issues. Second is the interaction with trusted health care providers. Third, the social support of family and others. Fourth, and perhaps most challenging, is adherence to both immunosuppressive and cystinosis related medications.

Kidney transplantation seems to be inevitable despite the best WBC cystine level control. However cysteamine treatment certainly delays the need for kidney transplantation in addition to reducing the other effects of cystinosis. The older age at transplant does mean that there is more time with chronic kidney disease. Managing the unique issues of a low GFR in someone with cystinosis requires continued potassium and phosphate supplementation; rarely required with severe chronic kidney disease due to other diseases.

For individuals with cystinosis, kidney transplantation may occur in later teenage years, in their twenties or even after 30 years of age. Early after transplantation, removal of the "native" kidneys may be beneficial given that ongoing loss of electrolytes and water due to continued "native" urine output threaten the crucial hydration and perfusion of the newly transplanted kidney. Without "native" nephrectomies supplementation that is required for continued native kidney electrolyte losses may have to be added to the magnesium and phosphate supplementation that is commonly needed in the first few months after transplantation.

The first few months after transplant can be difficult with adjustment to new medications including immunosuppressants, antimicrobial prophylaxis and high blood pressure medications. Immunosuppressive medications have adverse effects, particularly prednisone that increases appetite. Weight gain after kidney transplantation is common. While a good appetite may be welcome, especially after a period of poor food intake associated with low kidney function prior to transplantation, excess weight gain can be an issue. Obesity and medication adverse effects combined with the effects of cystinosis on pancreatic function make diabetes a possibility.

If a kidney transplant occurs before completion of growth, gaining height may be improved with the improved kidney function. If growth hormone was being used before transplantation its use is usually stopped but a restart may be considered, if needed, perhaps a year after a kidney transplant.

Adherence to medications after transplantation is important because immunosuppression is needed to optimize the health of the new kidney. There is an increased risk that teenagers/young adults, compared to children and older adults, do not take all their medications after kidney transplantation. This may explain why, in general, kidney transplants have a shorter "survival" time for teenagers. While some studies suggested that kidney transplants "survive" longer for individuals with cystinosis compared to other recipients, more recent studies show less of an advantage.

For some diseases that cause kidney failure recurrence in a transplanted kidney is a concern. Fortunately, this is not the case for cystinosis; the transplanted kidney is unaffected. However, while a kidney transplant replaces kidney function, it does not improve the many other cystinosis related issues. Thus cysteamine should, and can, be restarted as soon as possible after a kidney transplant.

"Quality of life" is undoubtedly improved by a kidney transplant. Social support from parents, the wider family, classmates and particularly others at school improve quality of life. There should be recognition of the potential for negative effects of worries such as how long the kidney transplant will function, future health and, in some systems, the costs of future healthcare. An important aspect of life after transplantation includes starting a family and there have been several reports of successful pregnancies after kidney transplantation for women with cystinosis.

Maintaining a connection to clinicians with expertise in cystinosis, in addition to the transplant team, may be necessary. This is a challenge when transitioning to a transplant centre (or to adult services). Transplant centres will be very familiar with care of a person with a new kidney, for example immunosuppression management and preventing infections, but not the other medical issues that individuals with cystinosis may have. Fortunately some issues that are often seen soon after kidney transplantation (such as hyperparathyroidism and hypertension) may be less frequent in individuals with cystinosis.

References:

1. A transition guide for teens and young adults with cystinosis and their families. https://cystinosis.org/wp-content/uploads/2019/01/FINAL_CRN_Transition_TeensAdults-3.pdf
2. Ariceta G et al. Cysteamine (Cystagon) adherence in patients with cystinosis in Spain: successful in children and a challenge in adolescents and adults. Nephrol Dial Transplant 17:1766 (2002) https://www.ncbi.nlm.nih.gov/pubmed/25348508
3. Gahl WA et al. Nephropathic cystinosis in adults: natural history and effects of oral cysteamine therapy Ann Int Med 147:242 (2007) https://www.ncbi.nlm.nih.gov/pubmed/17709758
4. Nesterova G and Gahl W. Nephropathic cystinosis: late complications of a multisystemic disease. Pediatric Nephrology 23:863 (2008) https://www.researchgate.net/publication/5839166_Nephropathic_cystinosis_Late_complications_of_a_multisystemic_disease
5. Beinart N et al. Mood and illness experiences of adults with cystinosis. Renal Failure 37:835 (2015) https://www.ncbi.nlm.nih.gov/pubmed/25715639
6. Cohen et al. Excellent long-term outcome of renal transplantation in cystinosis patients. Orphanet Journal of Rare Diseases. 10:90 (2015) https://ojrd.biomedcentral.com/articles/10.1186/s13023-015-0307-9
7. Doyle M et al. Family Strategies for Living with Rare Disease: The Experience of Cystinosis. Journal of the Society for Social Work and Research 7:547 (2016) https://cystinosis.org/wp-content/uploads/2019/01/DoyleWerner-Lin-2016-Family-Strategies-JSSWR.pdf
8. Berryhill A et al. Cysteamine in renal transplantation: A report of two patients with nephropathic cystinosis and the successful re-initiation of cysteamine therapy during the immediate post-transplant period. Pediatric Transplantation. 20:141-5 (2016). https://onlinelibrary.wiley.com/doi/10.1111/petr.12617
9. Raina R et al. Structured Transition Protocol for Children with Cystinosis. Frontiers in Pediatrics 5:191 (2017) https://www.researchgate.net/publication/319396751_Structured_Transition_Protocol_for_Children_with_Cystinosis
10. Kizilbash et al. Trends in kidney transplant outcomes in children and young adults with cystinosis. Pediatric Transplantation 23:e13572 (2019). https://onlinelibrary.wiley.com/doi/10.1111/petr.13572
11. Wightman A t al. Health-related quality of life changes following renal transplantation in children. Pediatric Transplantation. 23:e13333 (2019). https://orcid.org/0000-0003-0167-0420
12. Kasimer RN et al. Adult complications of nephropathic cystinosis: a systematic review. Pediatric Nephrology 36:223-236 (2021) https://cystinosis.org/wp-content/uploads/2020/02/Kasimer-2019-Adult-complications-in-cystinosis.Langman.pdf
13. Tucker E et at. Life and expectations post-kidney transplant: a qualitative analysis of patient responses. BMC Nephrology 20:175 (2019). https://bmcnephrol.biomedcentral.com/articles/10.1186/s12882-019-1368-0
14. Baskin E et al. Outcomes of renal transplantation in children with cystinosis. Transplantation 104:S554 (2020) https://doi.org/10.1097/01.tp.0000701520.16076.e4
15. Emma F et al. An international cohort study spanning five decades assessed outcomes of nephropathic cystinosis. Kidney International. 100:1112-1123 (2021) https://www.researchgate.net/publication/353028336_An_international_cohort_study_spanning_five_decades_assessed_outcomes_of_nephropathic_cystinosis
16. Reda A et al. Fertility in cystinosis. Cells 10:3539-3549 (2021). https://www.ncbi.nlm.nih.gov/pmc/articles/PMC8700558/
17. Achayra et al. Utility of post-transplant native nephrectomy in children with nephropathic cystinosis: A single centre retrospective study. Nephrology 28:469-471 (2013). https://onlinelibrary.wiley.com/doi/10.1111/nep.14189?af=R
18. Langman et al Fertility management in cystinosis: A clinical perspective. KI Reports 9:214-224 (2024) https://www.sciencedirect.com/science/article/pii/S2468024923015723
19. Roberts E et al. Family Environment, Social Support, and Health-Related Quality of Life in Adolescents Following Kidney Transplant. Pediatric Transplantation 28:e14824 (2024) https://onlinelibrary.wiley.com/doi/abs/10.1111/petr.14824
20. Alaskar M et al. Quality of Life in Pediatric CKD Patients on Dialysis vs Renal Transplantation: A Comparative Study of the Perspectives of Parents and Children. Transplantation Proceedings 57:241-249 (2025). https://www.sciencedirect.com/science/article/abs/pii/S0041134524006985