

CYSTINOSIS FUNDAMENTALS

MANAGEMENT CONSIDERATIONS FOR NEPHROLOGISTS

Nephropathic cystinosis is a rare, autosomal recessive lysosomal storage disorder¹⁻³



Inadequately treated cystinosis leads to multiorgan damage over time, which often progresses without apparent symptoms^{1,2,5-8}

Kidney transplant is not a cure. Following transplant, cystine continues to accumulate in extrarenal organs⁵

Patients with cystinosis are living longer with early diagnosis and advances in medical care^{1,5}

Cystinosis treatment includes^{2,9}:

- 1 Cystine-depleting therapy (CDT)
- 2 Management of renal disease and transplant
- 3 Medications for extrarenal manifestations

2 TYPES OF CDT are necessary for patients with nephropathic cystinosis, regardless of age or transplant status^{6,10}:



Cysteamine eye drops dissolve corneal crystals⁶



Oral (systemic) cysteamine reduces cystine levels in other cells and tissues⁶

- IR cysteamine (Q6H)¹¹
- DR cysteamine (Q12H)¹¹

Regular WBC cystine testing assesses disease control and CDT adherence and informs dose adjustments¹⁰⁻¹²



No-cost collection kits are available for WBC cystine level testing at WBCKit.com¹³



WBCKit.com

Target WBC cystine level depends on the type of test used^{11,13}

Early and continuous cystine control with CDT may prevent or limit damage^{6,14}

Sustained adherence to CDT may^{6,10,14-16}:

- Delay the need for kidney transplant until late teens or early adulthood
- Prolong kidney function; 1 year below target cystine is associated with an additional year of kidney function
- Reduce extrarenal complications
- Increase life expectancy



Nonadherence to CDT is common, and even brief delays can lead to significant increases in cystine levels^{17,18}

- Strict adherence can be difficult to maintain due to pill burden, dosing frequency, need for lifelong treatment, and adverse effects^{2,17,19}
- Treatment adherence and motivation to follow cystinosis treatment drastically decline starting in adolescence¹⁹

Routine monitoring is an essential part of cystinosis management^{2-4,9}

PARAMETER	FREQUENCY
WBC cystine levels	<ul style="list-style-type: none"> At least 4 times per year when at WBC cystine target (children) At least once or twice annually when at WBC cystine target (adults)
Chemistry profile, renal function, and graft health	As appropriate
Growth and development (children)	4 times per year
Endocrine, bone, and reproductive health	<ul style="list-style-type: none"> Every 6 months (if stable): <ul style="list-style-type: none"> Thyroid panel (TSH and T4) HbA1c Once per year (if stable): <ul style="list-style-type: none"> Bone: IGF-1, calcium, phosphorus, alkaline phosphatase, 25-hydroxy vitamin D, PTH Males: FSH, LH, testosterone, prolactin, inhibin B Females: FSH, LH, estradiol, anti-müllerian hormone, prolactin
Other (eg, neurocognitive assessments, psychological and mental health, treatment adherence, medication regimen review)	As appropriate

Coordinated multidisciplinary care is needed to address potential long-term complications^{1,4-7,11,20}

Monitor at Least TWICE ANNUALLY

Nephrologist/Transplant team



Cystinosis care coordination
Fanconi syndrome
ESRD
Rickets



Additional providers include:

Primary care provider; dietitian/nutritionist; geneticist; obstetrician-gynecologist; orthopedist; pharmacist; physical, occupational, or speech therapist; social worker; and urologist

Monitor at Least ONCE ANNUALLY

Ophthalmologist



Photophobia
Corneal and retinal complications

Neurologist/Neuromuscular specialist



Intracranial hypertension
Neurocognitive deficits
Myopathy and muscle wasting
Swallowing and breathing difficulty

Endocrinologist



Growth failure
Hypothyroidism
Male hypogonadism and infertility^a
Diabetes mellitus

Monitor AS APPROPRIATE

Mental health clinician



Neurocognitive deficits
Psychosocial or behavioral concerns

Gastroenterologist



Nausea/vomiting
Gastroesophageal reflux
Dysmotility
Feeding and swallowing difficulty
Hepatosplenomegaly

Pulmonologist



Breathing difficulty
Swallowing difficulty and aspiration

Cardiologist



Cardiomyopathy
Coronary artery calcification

Clinician and patient resources are available

For more information about cystinosis, please contact your Amgen representative.

Amgen Cystinosis Information for Clinicians



[UnderstandingCystinosis.com](https://www.wbckit.com)

Amgen Cystinosis Information for Patients



[CystinosisUnited.com](https://www.cystinosis.org)

Cystinosis Research Network



[Cystinosis.org](https://www.cystinosis.org)

Cystinosis Research Foundation



[CystinosisResearch.org](https://www.cystinosis.org)

^aCystinosis has not been shown to cause infertility in women.⁵

ESRD, end-stage renal disease; FSH, follicle-stimulating hormone; HbA1c, glycated hemoglobin; IGF-1, insulin-like growth factor-1; LH, luteinizing hormone; PTH, parathyroid hormone; T4, thyroxine; TSH, thyroid-stimulating hormone.

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