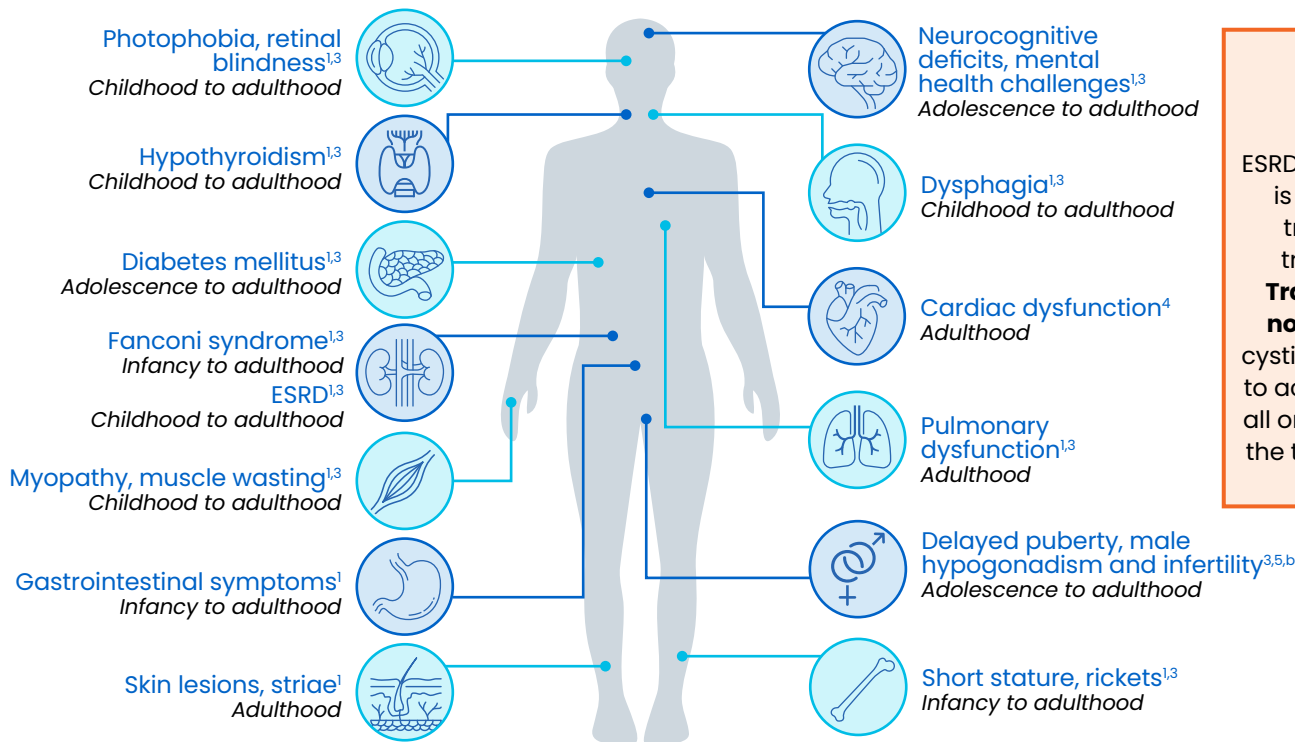


# CYSTINOSIS FUNDAMENTALS

## MULTIDISCIPLINARY MANAGEMENT CONSIDERATIONS FOR NEPHROLOGISTS

Cystinosis causes renal and extrarenal manifestations over time that require a multidisciplinary approach and treatment to improve long-term outcomes<sup>1,2,a</sup>



ESRD in cystinosis is ultimately treated by transplant. **Transplant is not a cure** as cystine continues to accumulate in all organs except the transplanted kidney.<sup>2</sup>

[UnderstandingCystinosis.com](https://www.understandingcystinosis.com) has information and resources for clinicians



Contact your [Amgen representative](#) about peer-to-peer educational programs with cystinosis experts

[CystinosisUnited.com](https://www.CystinosisUnited.com) has information and resources for patients and caregivers, including a [video](#) on multidisciplinary care



## Routine renal care and cystinosis management

### Recommended monitoring

- WBC cystine levels to monitor disease control and adherence and inform CDT dosage adjustment<sup>6</sup>
  - Children: at least 4x/year<sup>6,c,d</sup>
  - Adults: at least 1x/year<sup>6,c,d</sup>
- Preservation of native renal function in non-ESRD<sup>1,7</sup>
- Growth (children) and nutrition<sup>1,8</sup>
  - Height and weight
  - GH deficiency testing
  - Nutritional assessment
- TSH, HgbA1c monitoring (adults)<sup>1</sup>
- Chemistry profile and renal function<sup>1,4,8,9</sup>
  - Electrolytes, sCr, eGFR, uACR
  - Transplant: U/S (if needed) and immunosuppressant TDM

Request a no-cost WBC cystine level collection kit at [wbckit.com](https://www.wbckit.com)



### General check-in questions

- What symptoms have you been experiencing since your last appointment?
  - Is anything new or particularly concerning?
- How many doses of your CDT are you missing each week?
- What specialists have you seen since your last appointment?
  - Did any of them change your medication or recommend something new?
  - Are you having trouble scheduling any specialist appointments?

<sup>a</sup>Age ranges for image: infancy: 0 to 1 year; childhood: 2 to 12 years; adolescence: 13 to 18 years; adulthood: ≥19 years. <sup>b</sup>Cystinosis has not been shown to cause infertility in women.<sup>5</sup> <sup>c</sup>Target WBC cystine level for granulocyte test is <1.9 nmol ½ cystine/mg protein and <1.0 nmol ½ cystine/mg protein for mixed leukocytes.<sup>10</sup> <sup>d</sup>Patients with unstable or above-target WBC cystine levels may require more frequent testing. Refer to CDT prescribing information for recommended testing and follow-up.<sup>6</sup>

CDT, cystine-depleting therapy; eGFR, estimated glomerular filtration rate; ESRD, end-stage renal disease; GH, growth hormone; HgbA1c, glycated hemoglobin; sCr, serum creatinine; TDM, therapeutic drug monitoring; TSH, thyroid-stimulating hormone; uACR, urine albumin-creatinine ratio; U/S, ultrasound; WBC, white blood cell.

# Extrarenal cystinosis care—symptoms to look for and specialist referrals

Symptom probes Do you ever have...	Extrarenal manifestations	Nephrology clinic symptom testing	Specialist referral
<ul style="list-style-type: none"> <li>Light sensitivity?</li> <li>Trouble seeing at night?</li> <li>Headaches?</li> </ul>	<ul style="list-style-type: none"> <li>Photophobia<sup>8</sup></li> <li>Corneal and retinal complications<sup>4,8</sup></li> <li>Intracranial hypertension<sup>4,8</sup></li> </ul>	<ul style="list-style-type: none"> <li><b>Discuss</b> vision quality with patient<sup>4</sup></li> <li><b>Ask</b> about pain/discomfort around the eyes<sup>4</sup></li> <li><b>Order</b> brain MRI or CT<sup>1,4</sup></li> </ul>	<b>Ophthalmologist<sup>1,c:</sup></b> <ul style="list-style-type: none"> <li>Slit-lamp biomicroscopy<sup>1,8</sup></li> <li>Intraocular pressure<sup>1,8</sup></li> <li>Dilated fundus exam<sup>1,8</sup></li> </ul>
<ul style="list-style-type: none"> <li>Memory issues?</li> <li>Trouble gripping?</li> <li>Trouble swallowing?</li> <li>Trouble breathing?</li> <li>Anxiety? Depression?</li> <li>Trouble sleeping?</li> </ul>	<ul style="list-style-type: none"> <li>Neurocognitive deficits<sup>1,8</sup></li> <li>Myopathy and muscle wasting<sup>4</sup></li> <li>Swallowing difficulty<sup>4,8</sup></li> <li>Breathing difficulty<sup>4,8</sup></li> <li>Psychosocial or behavioral concerns<sup>1</sup></li> </ul>	<ul style="list-style-type: none"> <li><b>Perform</b> mini-mental status examination<sup>1</sup></li> <li><b>Assess</b> hand grip strength<sup>8</sup></li> <li><b>Discuss</b> swallowing challenges with patient<sup>4,8</sup></li> <li><b>Measure</b> O<sub>2</sub> saturation and blood gas<sup>8</sup></li> <li><b>Conduct</b> psychological screening inventory<sup>1</sup></li> <li><b>Order</b> sleep study<sup>1</sup></li> </ul>	<b>Neurologist<sup>1,b;</sup> physical/occupational therapist<sup>1,c;</sup> SLP<sup>1,c:</sup></b> <ul style="list-style-type: none"> <li>Electromyography<sup>4</sup></li> <li>Musculoskeletal examination<sup>8</sup></li> <li>Muscular biopsy<sup>8</sup></li> <li>Swallowing test<sup>4</sup></li> </ul> <b>Pulmonologist<sup>8,b:</sup></b> <ul style="list-style-type: none"> <li>Pulmonary function tests</li> <li>Spirometry</li> </ul> <b>Mental health clinician<sup>1,c;</sup> Social worker<sup>1,c:</sup></b> <ul style="list-style-type: none"> <li>Psychological assessment</li> </ul>
<ul style="list-style-type: none"> <li>Bone aches?</li> <li>Trouble with the cold?</li> <li>Frequent thirst or urination?</li> <li>Desire to father children?</li> </ul>	<ul style="list-style-type: none"> <li>Growth failure<sup>1,11</sup></li> <li>Rickets and orthopedic challenges<sup>1,11</sup></li> <li>Hypothyroidism<sup>1</sup></li> <li>Diabetes mellitus<sup>1</sup></li> <li>Male hypogonadism and infertility<sup>1</sup></li> </ul>	<ul style="list-style-type: none"> <li><b>Check</b> GH<sup>11</sup></li> <li><b>X-ray</b> for bone deformities<sup>11</sup></li> <li><b>Assess</b> bone health (serum iPTH, Ca, PO<sub>4</sub>, ALP, and bicarbonate levels)<sup>11</sup></li> <li><b>Test</b> thyroid function (FT3, T4, TSH)<sup>1</sup></li> <li><b>Measure</b> fasting glucose and HgbA1c<sup>1</sup></li> <li><b>Assess</b> gonad functioning (testosterone, inhibin B, LH, FSH)<sup>1</sup></li> </ul>	<b>Orthopedist<sup>1,c</sup></b> <ul style="list-style-type: none"> <li>Bone biopsy<sup>11</sup></li> </ul> <b>Endocrinologist<sup>1,b</sup></b>
<ul style="list-style-type: none"> <li>Upset stomach or vomiting?</li> <li>Heartburn?</li> <li>Constipation or diarrhea?</li> <li>Upper right abdominal pain?</li> <li>Trouble eating?</li> </ul>	<ul style="list-style-type: none"> <li>Nausea/vomiting<sup>12</sup></li> <li>Gastroesophageal reflux<sup>12</sup></li> <li>Dysmotility<sup>12</sup></li> <li>Hepatosplenomegaly<sup>12</sup></li> <li>Feeding difficulty<sup>12</sup></li> </ul>	<ul style="list-style-type: none"> <li><b>Review</b> GI symptom history<sup>12</sup></li> <li><b>Run</b> liver function tests (ALT, AST, GGT, ALP, total protein, albumin, bilirubin, and PT)<sup>12,13</sup></li> <li><b>Monitor</b> nutritional status<sup>1,12</sup></li> </ul>	<b>Gastroenterologist<sup>1,12,b:</sup></b> <ul style="list-style-type: none"> <li>Endoscopy</li> </ul>
<ul style="list-style-type: none"> <li>High blood pressure?</li> <li>Chest pain? Rapid heartbeat?</li> <li>High cholesterol?</li> </ul>	<ul style="list-style-type: none"> <li>Hypertension<sup>1,9</sup></li> <li>Cardiomyopathy<sup>4,9</sup></li> <li>Dyslipidemia<sup>8</sup></li> </ul>	<ul style="list-style-type: none"> <li><b>Measure</b> blood pressure<sup>1,4</sup></li> <li><b>Check</b> lipid profile (total cholesterol, LDL, HDL, and TG)<sup>8,15</sup></li> </ul>	<b>Cardiologist<sup>1,b</sup></b> <ul style="list-style-type: none"> <li>Electrocardiogram<sup>8</sup></li> <li>Echocardiogram<sup>14</sup></li> </ul>

Additional providers to include as appropriate: primary care provider<sup>5;</sup> dermatologist<sup>1;</sup> dentist<sup>1;</sup> geneticist/genetic counselor<sup>15;</sup> obstetrician-gynecologist<sup>5;</sup> and pharmacist.<sup>12</sup>

<sup>a</sup>Follow-up recommended at least every 6 to 12 months.<sup>1</sup> <sup>b</sup>Follow-up recommended at least annually.<sup>1,8</sup> <sup>c</sup>Follow-up recommended as appropriate.<sup>1</sup>

ALP, alkaline phosphatase; ALT, alanine transaminase; AST, aspartate aminotransferase; Ca, calcium; CT, computed tomography; FSH, follicle-stimulating hormone; FT3, free triiodothyronine; GGT, gamma-glutamyl transferase; GI, gastrointestinal; HDL, high-density lipoprotein; iPTH, intact parathyroid hormone; LDL, low-density lipoprotein; LH, luteinizing hormone; MRI, magnetic resonance imaging; O<sub>2</sub>, oxygen; PO<sub>4</sub>, phosphate; PT, prothrombin time; SLP, speech language pathologist; T4, thyroxine; TG, triglycerides.

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