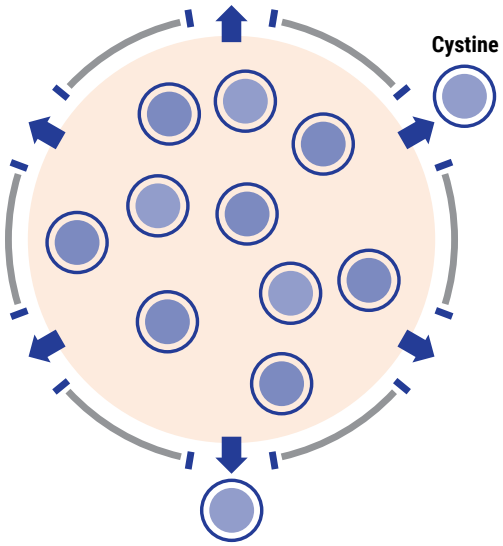
Know Cystinosis

What Is Cystinosis?

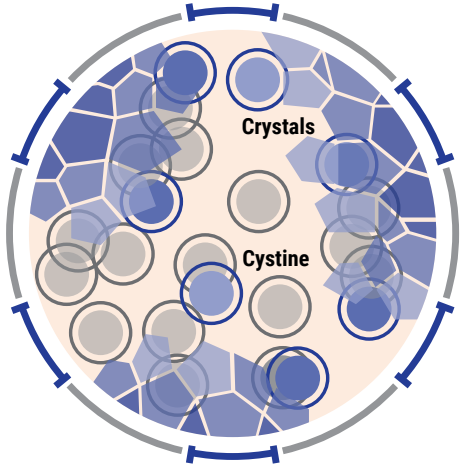
Nephropathic cystinosis is a rare, inherited, metabolic disorder that affects about 500 to 600 people in the United States.[1]

It is a lysosomal storage disorder (LSD) that results in the amino acid cystine accumulating inside the lysosomes of nearly every cell in the body. Cystine accumulation results in the formulation of crystals that lead to cell damage and death in tissues and organs throughout the body.[2]

Lysosome in a person without cystinosis



Lysosome in a person with cystinosis



Early Signs and Symptoms of Cystinosis

- Initial symptoms are commonly the result of cystine accumulation in the kidney tubules, which results in Fanconi syndrome. If untreated, Fanconi syndrome will lead to end-stage renal disease, requiring a kidney transplant by age 10.[2]
- Parents primarily note frequent wet diapers (polyuria) and persistent thirst (polydipsia, resulting in dehydration) when first describing their child's symptoms.[2]
- Also commonly affected in the first 6 to 18 months of life are the eyes, which can become photosensitive, and the bones, which can develop rickets. Damage to the thyroid further results in failure to thrive.[2]

For more about cystinosis, please contact your Horizon Clinical Science Associate.

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Signs of Elevated Cystine Levels Can Be Seen in Nearly Every Cell of the Body[2,3]

Damage occurring in the body is most readily observed in the kidneys. While a transplant is often needed to restore function, it may be delayed until early adulthood with early and consistent cystine-depleting therapy (CDT).[2,3]

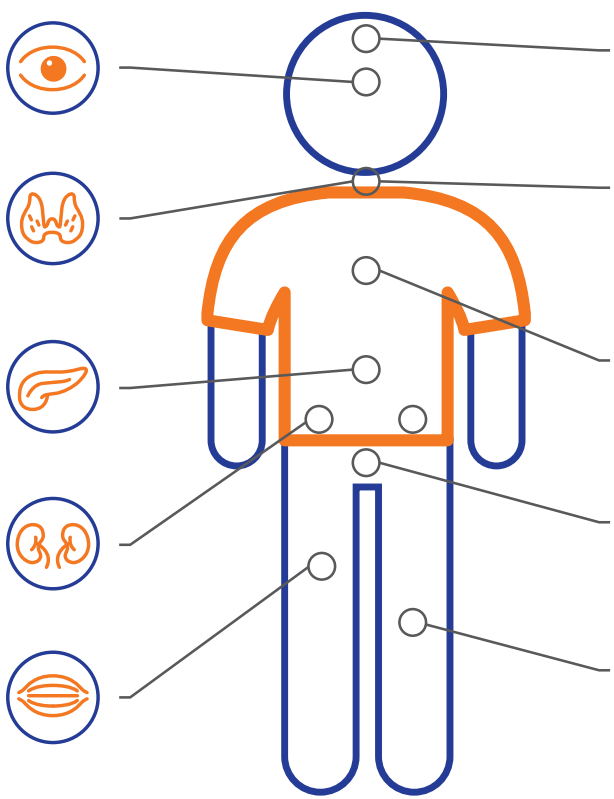
Eyes
Sensitivity to light (photophobia)
childhood and adulthood
Blindness
childhood and adulthood

Thyroid
Poorly working thyroid (hypothyroidism)
childhood and adulthood

Pancreas
Diabetes
childhood and adulthood

Kidneys
Fanconi syndrome
infancy, childhood, and adulthood
Kidney failure
childhood and adulthood

Muscles
Muscle weakness and decreased muscle mass (myopathy)
childhood and adulthood



Brain
Visual or learning issues
childhood and adulthood

Throat
Trouble swallowing
childhood and adulthood

Lungs
Problems breathing
adulthood

Male reproductive organs*
Not being able to father children naturally (infertility)
adulthood

Bones
Softening or weakening or bones (rickets)
infancy, childhood, and adulthood

*Cystinosis has not been shown to cause infertility in women.

Cystinosis Is Progressive but Manageable

With cystine-depleting therapy (CDT), cystine levels may be controlled and some damage to organs may be prevented or limited.[4,5]

Support for Patients

Encourage patients to learn more about cystinosis, discover advocacy programs, and connect with others who are living with cystinosis.

Cystinosis® United

Cystinosis United is a resource from Horizon Therapeutics for people living with or caring for someone with cystinosis, offering helpful tips, support, and more.

Cystinosisunited.com

For more about cystinosis, please contact your Horizon Clinical Science Associate.

References:
1. Nesterova G, Gahl WA. Infantile nephropathic cystinosis standards of care — a reference for people with infantile nephropathic cystinosis, their families, and medical team. *Cystinosis Research Network*. June 2012. Accessed July 23, 2021. https://www.cystinosis.org/publications/infantile-nephropathic-cystinosis-standards-of-care/ **2.** Veys KRP, Besouw MTP, Pinxten A, et al. Cystinosis: a new perspective. *Acta Clinica Belgica*. 2016;71(3):131-137. **3.** Nesterova G, Gahl W. Nephropathic cystinosis: late complications of a multisystemic disease. *Pediatr Nephrol*. 2008;23(6):863-878. **4.** Nesterova G, Williams C, Bernardini I, et al. Cystinosis: renal glomerular and renal tubular function in relation to compliance with cystine-depleting therapy. *Pediatr Nephrol*. 2015;30:945-951. **5.** Langman CB, Barshop BA, Deschênes G, et al. Controversies and research agenda in nephropathic cystinosis: conclusions from a "Kidney Disease: Improving Global Outcomes" (KDIGO) Controversies Conference. *Kidney Int*. 2016;89(6):1192-1203.

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