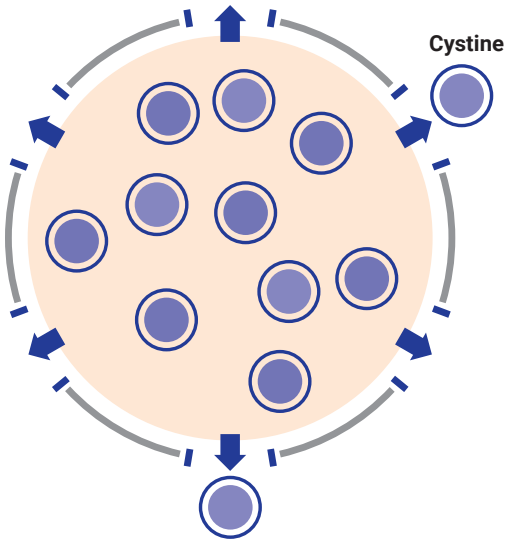
Know Cystinosis

What Is Cystinosis?

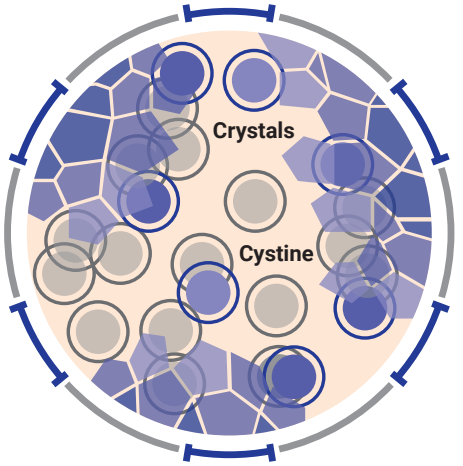
Nephropathic cystinosis is a rare inherited, metabolic disorder that affects approximately 100 people in Canada.[1]

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]

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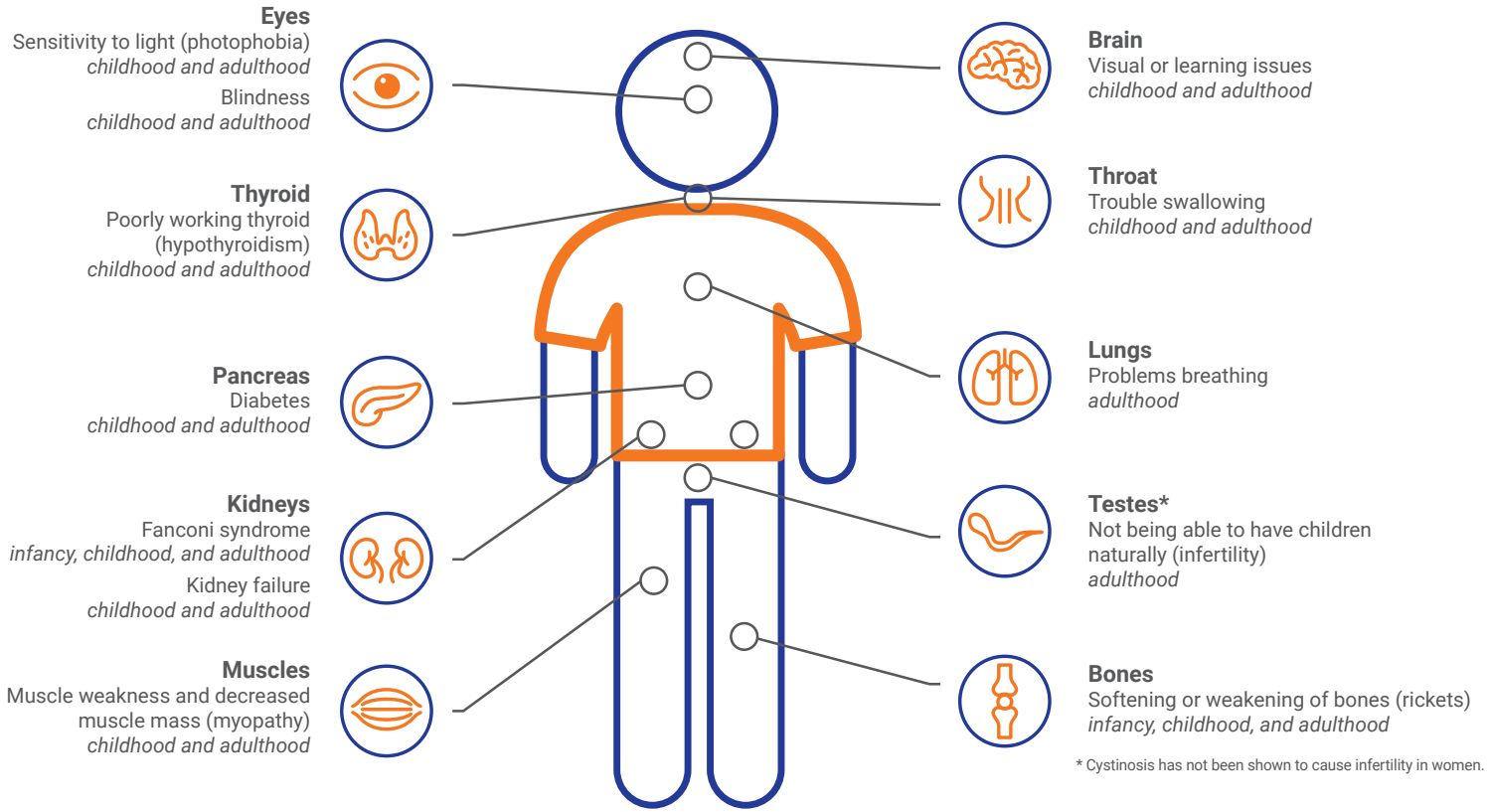
Signs of Elevated Cystine Levels Can Be Seen in Nearly Every Cell of the Body[2,3]

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 an early and consistent treatment regimen.[2,3]



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

Cystinosis is Progressive but Manageable

With appropriate treatment, 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

References:
1. Casey B. CANADIANS AFFECTED BY RARE DISEASES AND DISORDERS: IMPROVING ACCESS TO TREATMENT. Report of the Standing Committee on Health. The House Of Commons Canada. 2019;42(1):1-44. **2.** Veys KRP, 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, 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, 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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