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# Cystinosis

National Cancer Institute

## Source

National Cancer Institute. *Cystinosis*. NCI Thesaurus. Code C2976.

An autosomal recessive hereditary disorder characterized by defective transportation of cystine across the lysosomal membranes and systemic deposition of cystine crystals in the body. It is associated with slight increase of the plasma cystine, cystinuria, aminoaciduria, glycosuria, polyuria, hypophosphatemia, rickets, and renal tubular dysfunction.