Cystinuria

National Cancer Institute

Source

National Cancer Institute. Cystinuria. NCI Thesaurus. Code C84664.

An autosomal recessive inherited metabolic disorder caused by mutations in the SLC3A1 and SLC7A9 genes. It is characterized by deficient re-absorption of cystine in the proximal tubules of the kidney. It results in the formation of stones in the kidney, ureter, and urinary bladder.