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Cystinosis

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Cystinosis.

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Cystinosis is a metabolic disease characterized by an accumulation of cystine inside the lysosomes, causing damage in different organs and tissues, particularly in the kidneys and eyes. Three clinical forms have been described: nephropathic infantile, nephropathic juvenile and ocular (see these terms).