

Cystinuria

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CYSTINURIA

- cysteine (cys) is an amino acid derived from the breakdown of proteins in the lysosome (cell compartment)
- cystine (cys-cys) is formed by joining 2 cysteines
 $\text{cys-SH} + \text{HS-cys} \rightarrow \text{cys-S-S-cys}$
- defect = protein that prevents cystine (and certain other amino acids) from being reabsorbed into the kidney epithelial cells (see Figure 1)
- cystine accumulates in the urine and because of poor solubility in water leads to formation of crystals in the urinary tract and in the kidney tubules, the latter leading possibly to severe kidney damage.

