

FANCONI SYNDROME



PATHOPHYSIOLOGY:

Global dysfunction of the proximal tubule dysfunction as a result of genetic disease (eg. cystinosis, glycogen storage disease), drugs (eg. cisplatin, tenofovir) or systemic disease (eg. multiple myeloma, Sjögren's syndrome).

RESULT:

Since the proximal tubule is responsible for the reabsorption of several organic and inorganic compounds, Fanconi Syndrome results in urinary wasting of several compounds, including glucose, phosphorus, uric acid, amino acids and HCO_3^- . A metabolic acidosis can also develop due to reduced HCO_3^- reabsorption (see proximal RTA).