

FANCONI SYNDROME & THE PROXIMAL TUBULE

Proximal Tubule Physiology

FANCONI SYNDROME

Proximal Tubule
MELT DOWN

DIAGNOSIS

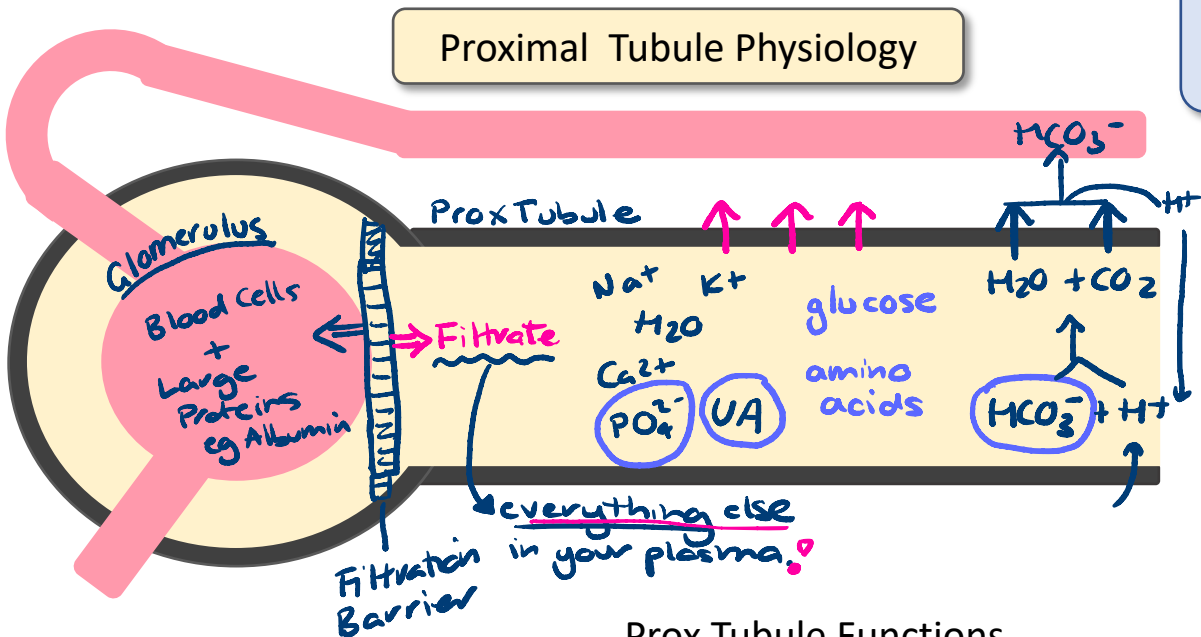
AETIOLOGY

ACQUIRED

HEREDITARY

Prox Tubule Functions

FANCONI SYNDROME & THE PROXIMAL TUBULE



Prox Tubule Functions

MASS RECYCLING (Reabsorption) of Nutrients + Electrolytes

Acid / Base

Vit D activation

Recycling Small proteins eg Light Chains

FANCONI SYNDROME

Proximal Tubule MELT DOWN

RTA (Renal Tubular Acidosis) proximal type II

- ↓ HCO₃⁻
- ↑ Cl⁻
- normal anion gap
- ↓ K⁺

low serum phosphate

low serum uric acid.

DIAGNOSIS

glycosuria
amino aciduria

Tubular Proteinuria

↑ Proteinuria but not Albuminuria i.e PCR > ACR

AETIOLOGY

ACQUIRED

Drugs eg tenofovir, toluene
Heavy metals
Myeloma
Autoimmune

HEREDITARY

inborn errors of metabolism eg Wilson's Disease, Fructose intolerance + others