Fanconi Syndrome Characteristics

Fanconi syndrome is a disease of the kidney where a resorption defect in the proximal tubule causes various substrates and electrolytes to be excreted in the urine, such as amino acids, glucose, bicarbonate, and phosphate. As a result, patients suffer electrolyte deficiencies and their sequelae. Treatment of Fanconi syndrome starts with treating the underlying disorder and replacing the lost electrolytes.



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Pathophysiology

Resorption Defect in Proximal Tubule
Not Absorbing Electrolytes in P Tube
Fanconi Syndrome is characterized by a resorption defect in the proximal tubule leading to an increased excretion of phosphate, potassium, and bicarbonate from the urine.

Signs and Symptoms

Hypophosphatemia
Hippo-phosphate-P
Patients with Fanconi syndrome have hypophosphatemia due to an increase in loss of phosphate in the defective proximal tubule. This can lead to rickets in children and in adults.

Rickets
Racket of bone
A condition causes by decreased mineralization of bones. It can be due to low vitamin D, calcium, or phosphate. This term has been used to describe the condition in children whereas osteomalacia is the term for adults.

Osteomalacia
Ostrich-Malaysia
A condition caused by the decreased mineralization of bones due to low levels of calcium or phosphate. This term is typically used to describe the condition in adults whereas rickets is the term commonly used for children.

Hypokalemia
Hippo-banana
Due to the dysfunction of the proximal convoluted tubule, sodium reabsorption is diminished. This sodium loss contributes to an environment of hypovolemia and secondary hyperaldosteronism, which are coupled with the increased distal delivery of sodium to encourage sodium reabsorption and potassium secretion in the collecting tubule, resulting in hypokalemia.

Metabolic Acidosis
Metal-ball Acidic-lemon
In Fanconi syndrome, excess loss of bicarbonate causes patients to develop metabolic acidosis.

Type 2 Renal Tubular Acidosis
(2) Tutu Kidney Tuba with Acidic-lemon
In Fanconi syndrome, patients develop metabolic acidosis due to excess bicarbonate loss, and this is classified specifically as type 2 renal tubular acidosis.

Treatment

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Treat Underlying Disorder

Underlying Diseased-guy

Managing Fanconi syndrome starts with treating the underlying disorder, in other words, stopping the excess electrolyte excretion. There are many disorders that can cause Fanconi syndrome, including hereditary vs non-hereditary etiologies. Some of these disorders can be reversed.

Bicarbonate

Bi-car-bomb

After addressing the underlying disorder, lost bicarbonate can be replaced to treat the metabolic acidosis.

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