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Acid-Base Balance-Electrolyte Quiz – Case 50

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A 16-year-old girl was presented with muscle weakness due to hypokalemia (serum potassium 2.6 mEq/L). Biochemical evaluation revealed serum glucose 90 mg/dL, creatinine 0.7 mg/dL, sodium 140 mEq/L, chloride 112 mEq/L, calcium 9.8 mg/dL, and phosphate 2.8 mg/dL. Arterial blood gas analysis showed arterial pH 7.33, PCO_2 35 mmHg and bicarbonate 18 mEq/L. Urine examination revealed glucosuria (2+).

All the below stated tests are necessary for the diagnosis of the underlying disease, except from:

a. Determination of serum ceruloplasmin levels among with ophthalmological examination

b. Determination of autoantibodies [ANA, anti-Ro-(SSA), anti-La (SSB)]

c. Protein electrophoresis, immunoelectrophoresis in both serum and urine, as well as determination of immunoglobulins

d. U/S examination of the kidneys

Comment

The patient developed a rather generalized abnormality of the proximal tubular function (Fanconi syndrome) associated with hypokalemia due to renal potassium wasting (urine potassium was 45 mEq/g creatinine), hypophosphatemia due to inappropriate phosphaturia ($FEPO_4^{3-}$ was 36%), renal glucosuria, and hyperchloremic metabolic acidosis with a normal anion gap (10 mEq/L) (proximal renal tubular acidosis). Monoclonal gammopathy and autoimmune diseases (such as Sjögren syndrome) are the most common causes among adults; thus, protein electrophoresis, immunoelectrophoresis in both serum and urine, and the determination of autoantibodies are necessary in the diagnostic workup of patients with Fanconi syndrome. On the other hand in children and adolescents, metabolic diseases, such as Wilson's disease are common causes. Thus, the determination of serum cerulospasmin levels and a slit-lamp examination of the eyes are especially useful for the diagnosis of Wilson's disease. In fact, in the present case, serum cerulospasmin levels were low and the slit-lamp examination revealed the presence of the Kayser-Fleischer ring, findings indicative of the disease.

Wilson's disease is an inherited disease characterized by a defect of copper transport by the liver lysosomes, resulting in excess deposition of copper in various tissues, including the renal tubules. Interestingly, refractory rickets due to Fanconi syndrome secondary to Wilson's disease has been occasionally described.

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Diagnosis: *U/S examination of the kidneys*