Author's response to reviews

Title: De Toni-Debre-Fanconi Syndrome in a patient with Kearns-Sayre Syndrome: a case report

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Dear Editors,

Thank you very much for your guidance! We revised the article and we answered below to your comments:

The authors mention in the paper hyperaldosteronism and hypoparathyroidism. Was the hyperaldosteronism secondary, as renin also seems to be elevated?

Usually, a drop of serum potassium by 0.3 mmol can reduce aldosterone secretion significantly, up to 45%!

ANSWER: The combination: hypotension/normotension + high serum renin + high serum aldosterone => suggests: secondary hyperaldosteronism. Decreased Na reabsorption in the proximal tubule, as a result of acidosis itself, leads to increased Na delivery to the distal tubule and promotes K secretion. Increased sodium loss and volume depletion leads to secondary hyperaldosteronism. Increased bicarbonate delivery to the distal tubule leads to increased K loss. Diminished distal tubular Na reabsorption reduces the negative intraluminal gradient and as a result, decreases both H and K secretion. The consequence is an increased Na, K and Ca loss in the urine and clinically: nephrolithiasis, osteomalacia, muscle weakness, growth retardation in children and renal insufficiency. The presence of Fanconi syndrome is characteristic in association with proximal renal tubular acidosis. Our patient was diagnosed in a different hospital with renal tubular dysfunction, with decreased urine-concentrating ability, and excessive excretion of potassium and magnesium. The renal dysfunction was thought to have resulted from Bartter syndrome, due to the presence of hyperaldosteronism and hypokaliemia.

Also, in patients with hypoparathyroidism, Fahr syndrome, that is calcification of the basal ganglia can occur. I do not see this feature in the attached CT figures and think the CT figures could be deleted, as they do not show anything special.

ANSWER: We deleted the CT figures, as you suggested. Also, concerning this
association: KSS and hypoparathyroidism we detailed in this presentation the signs and symptoms related to hypocalcemia, as well as the laboratory results that confirmed the hypoparathyroidism.

In regards to the diabetes of this patient, with progressive renal failure, why did the authors not choose regular insulin instead of insulin analogue?

ANSWER: Progressive renal failure-induced metabolic disorders should be addressed when treating patients with diabetes, to ensure the introduction of adequate therapy adjustments with the onset of renal function decline. The action of regular insulin may be prolonged as a consequence of the failure of renal insulin degradation, making the dose-effect profile of insulin difficult to control, and hypoglycemia more likely.

Managing diabetes in patients with end-stage renal disease is often problematic, because renal failure interferes with the metabolism of glucose and insulin with wide fluctuations in their daily blood glucose profile.

There is evidence that using insulin analogues in patients with diabetes and renal failure is safe and help to avoid large fluctuations in blood glucose levels (Jehle et al., 1999); also, insulin aspart in renal impaired patients has shown unaltered pharmacokinetics and safety profile in persons with diabetes with various degrees of renal dysfunction (Lyness W, Tyler JF, Lawrence A. Renal Impairment does not affect Insulin Aspart pharmacokinetics in type 1 diabetes. Diabetes 2001; 50 (2): A44).

I suggest to read and cite the following papers:


Isotani H et al., Hypoparathyroidism and insulin-dependent diabetes....Clin Endocrinol 1996

Dewhurst AG et al., Kearns Sayre Syndrome, hypoparathyroidism, and basal ganglia calcification.

ANSWER: Thank you very much for you suggestions! We read these articles and cited in our case presentation.