Reviewer’s report

Title: The Clinical Significance of Aldosterone Synthase Deficiency: Report of a Novel Mutation in the CYP11B2 gene

Version: 1 Date: 22 February 2014

Reviewer: Elise P Gomez-Sanchez

Reviewer’s report:

All minor issues: correction will enhance clarity.

Suggested changes for syntax:
Abstract:
“…facilitate clinical management of the patient and ASSESSMENT OF the genetic implication in their offspring”

Last sentence of discussion: “NEITHER mutation WAS detected in the patient’s brother.”

4th paragraph of the conclusions: Only indapamide? Or any thiazide or thiazide-like diuretic?

Please clarify these 2 sentences. Was this the same patient?
“Generalised weakness with marked hyperkalaemia and dehydration after institution of indapamide for hypertension and bowel preparation for barium enema has been reported as the first presentation in a middle-aged man[13]. Interestingly, past history revealed vomiting and failure to thrive in early infancy that resolved without any mineralocorticoid replacement[13].”

This still happens, so should be in present tense:
“It has been shown that mineralocorticoid receptor expression in human kidneys began with low levels in late gestation and rose progressively after birth[14]”

Level of interest: An article of outstanding merit and interest in its field

Quality of written English: Acceptable

Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:
I have no competing interests with the manuscript or authors.