Reviewer's report

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

Version: 1
Date: 24 February 2014
Reviewer: Michael Stowasser

Reviewer's report:

This is a well written paper describing a novel mutation in CYP11B2 causing aldosterone synthase deficiency. The following comments are minor and mostly discretionary only and aimed at improving the paper.

1. Page 2 - The background section of the Abstract is rather long and could be shortened. Suggest delete the first sentence as it is rather common knowledge.

2. Page 2, line 12 – suggest insert “Chinese” after “33-year old”.

3. Page 2, line 17 – suggest insert “CYP11B2” after “novel”.

4. Page 2, lines 22-24 – suggest reword to “Although a rare cause of hyperreninaemic hypoaldosteronism, aldosterone synthase deficiency should be suspected and the diagnosis sought in patients who present with life-threatening salt-wasting in infancy, as it has a good … etc”.

5. Page 4, line 2 – suggest replace “electrolyte” with “sodium” to be more specific.

6. Page 4, lines 5 and 6 – as small amounts of CYP11B2 expression have been reported in tissues other than the adrenal cortex, suggest reword to “It is synthesized by aldosterone synthase, an enzyme encoded by CYP11B2, expression of which is almost entirely confined to the adrenal cortex and exclusively in the zona glomerulosa layer.”

7. Page 4, line 15 – suggest insert “individuals” after “Indian”.


9. Page 4, line 28 – insert “at the time of this report” after “The index patient, “.

10. Page 5, line 2 – change “has” to “had”.

11. Page 5, line 13 – insert “years” after “33”.

12. Page 6, line 7 – insert “in whom” after “hyperreninaemic hypoaldosteronism, and “.

13. Page 6, line 17 – replace “until further evaluation with” with “pending”.

14. Page 7, line 3 – insert “and” after “of 18OHB, “.

15. Page 7, line 22 – replace “began” with “begins” and in line 23 replace “rose” with “rises”.

16. Page 8, line 1 – delete “the” and in line 2 change “more” to “greater”.

17. Page 8, line 5 – insert “therefore” after “may”.

18. Page 9, line 9 – insert “the” before “secondary structure”.

19. Page 9, line 12 – “and” after “activity (16.3%), “.

Level of interest: An article of importance in its field

Quality of written English: Needs some language corrections before being published

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

Declaration of competing interests:

I declare that I have no competing interests.