Reviewer's report

Title: Autosomal dominant pseudohypoaldosteronism type 1 with a novel splice site mutation in MR gene

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Reviewer: Gilberta Giancchetti

Reviewer's report:

In this paper “Autosomal dominant pseudohypoaldosteronism type 1 with a novel splice mutation in MR gene” describes a novel MR mutation in a patient with PHA. This is a well written paper. However there are several limitations that need to be addressed.

The background is poor and the authors could add more information about the classification of the pseudohypoaldosteronism and about the genetic alterations of this disease.

Is not really clear the relationship between mutation and phenotype. Are sure the authors that the genetic alteration involved only the MR receptor and not other genes? Discuss this point.

Recent papers have been published on this topic.

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

Quality of written English: Acceptable

Statistical review: Yes, and I have assessed the statistics in my report.