Reviewer's report

Title: An unexpected, mild phenotype of glucocorticoid resistance associated with glucocorticoid receptor gene mutation Case report and review of the literature

Version: 1 Date: 27 Jul 2017

Reviewer: David W. Ray

Reviewer's report:

The paper describes the expert evaluation of a patient with infertility which resulted in the discovery of glucocorticoid resistance. A glucocorticoid receptor mutation was discovered which may plausibly explain the phenotype. The mutation has been described before. A sibling also carries the mutation, but has no phenotype, therefore, it is possible that the mutation is not causative, but a polymorphism. The function of the mutant receptor was not tested in this paper.

A mild/negligible phenotype with GR allele loss was reported before...Trebble et al JCEM, and this should be referenced.

The introduction contains very many errors of grammar, and science, especially in relation to the GR structure and function.

Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

Yes

Does the work include the necessary controls?
If not, please specify which controls are required in your comments to the authors.

Yes

Are the conclusions drawn adequately supported by the data shown?
If not, please explain in your comments to the authors.

Yes
Are you able to assess any statistics in the manuscript or would you recommend an additional statistical review?
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Not relevant to this manuscript

Quality of written English
Please indicate the quality of language in the manuscript:

Not suitable for publication unless extensively edited

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