Reviewer's report


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Reviewer: Klaus Brusgaard

Reviewer's report:
The manuscript is interesting taking into account the extended use of methods to understand the phenotype of the proband. Even so a number of points needs to be addressed.
1. Figure 1 is of a very low quality.
2. It is stated that more than CYP11B1 100 mutations are known using HGMD I see 148. Thus, 100 is not precise enough.
3. The nomenclature of the chimeric variant to my knowledge does not follow common mutation nomenclature.
4. The methods used to explain the findings are not explained ie. which machines, what reagents, which softwares fx. the bioinformatics and molecular modeling? and so on.
5. A few numbers are jumping in supplementary table 1
6. In order to prove that the two mutations are not placed in cis sequencinig data from the father is needed - or other methods that can show independent segregation of the two variants.
7. A statistic of proband NGS coverage relative to a normal material does not say anything about the sequencing quality ie. reading depht or on target coverage.
8. The method used to derive quantitative data from the NGS data are not addressed.

Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.
No

Does the work include the necessary controls?
If not, please specify which controls are required in your comments to the authors.
No
Are the conclusions drawn adequately supported by the data shown?
If not, please explain in your comments to the authors.
No

Are you able to assess any statistics in the manuscript or would you recommend an additional statistical review?
If an additional statistical review is recommended, please specify what aspects require further assessment in your comments to the editors.
Not relevant to this manuscript

Quality of written English
Please indicate the quality of language in the manuscript:
Needs some language corrections before being published

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