Reviewer's report

Title: SLC5A2 mutations, including two novel mutations, responsible for renal glucosuria in Chinese families

Version: 2 Date: 28 Oct 2019

Reviewer: Lambertus P. W. J. van den Heuvel

Reviewer's report:

The manuscript of Yu et al. entitled "SLC5A2 mutations including two novel mutations responsible for Chinese renal glucosuria families (reference BNEP-D-19-00559R2)" reports a mutational detection study in another 10 Chinese families with FRG. In the majority of the families one heterozygous mutation has been established. In general in these families mild glucosuria is observed. This is an interesting manuscript which still can be improved. I suggest next issues will be addressed:

1.) Table 1: Under glucose excretion the code "-" means not tested or not present. Please add this information in the legend.

2.) Are allele frequencies for the observed variants in the Chinese population (like ExAc and gnomAD) known? If so, please add this information.

3.) Did the authors of the current manuscript establish the effect of the observed splice site mutations in the lymphoblastoid cell lines. If yes, please add these data.

4.) Did you check for the presence of the observed splice site mutations in every family of this manuscript (see table 1)?

5.) Why not develop (novel) specific primers to check for the presence of the observed splice site mutations on genomic DNA?
6.) Can you perform mutation prediction effect for the missense mutations with SIFT and Align (or some additional software programs)?

7.) Please check the manuscript for typo's.

Advice: Minor revision.

**Please confirm that you have included your review in the ‘Comments to Author’ box?**

As a minimum standard, please include a few sentences that outline what you think are the authors' hypothesis/objectives, their main results, and the conclusions drawn. Your report should constructively instruct authors on how they can strengthen their paper to the point where it may be acceptable for publication, or provide detailed reasons as to why the manuscript does not fulfill our criteria for consideration. Please supply appropriate evidence using examples from the manuscript to substantiate your comments. Please break your comments into two bulleted or numbered sections: major and minor comments.

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**Are the methods appropriate and well described to allow independent reproduction of experiments?**

Please state in the ‘Comments to Authors’ box below what you think are the strengths and weaknesses of the methods (study design, data collection, and data analysis), and what is required, if anything, to improve the quality of reporting.

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No

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- Are the statistical test(s) used in this study appropriate and well described?
- Is the exact sample size (n) reported for each experimental group/condition (as a number, not a range)?
- Are the description of any error bars and probability values appropriate?
- Are all error bars defined in the corresponding figure legends?
- Has a sample size calculation been included, or a description and rationale about how sample sizes were chosen?

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