Author's response to reviews

Title: Ethnic disparity in 21-hydroxylase gene mutations identified in Pakistani congenital adrenal hyperplasia patients

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Author's response to reviews:

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Robin Cassady-Cain, PhD
on behalf of:
Adrian Aldcroft, BA(Hons)
Executive Editor
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BioMed Central

Dear Sir,

Re: MS: 8842113214513104 - Ethnic disparity in 21-hydroxylase gene mutations identified in Pakistani congenital adrenal hyperplasia patients

With reference to the above mentioned manuscript. Please note that appropriate changes have been made according to the reviewer’s comment as follows:

Comments from Reviewer Leonidas Phylactouu

Major revisions

1 : The reviewer point is valid. But at present, it is not possible to perform additional studies due to lack of resources. However, we hope to establish the method in future. This has been addressed in discussion

2: Discussion has been modified and restructured

3: Figure 1 of the amplified fragments has been added
Minor revisions
1: The female to male ratio has been corrected
2: It has been specified in the introduction about inclusion of patients in previously published papers
3: The manuscript has been corrected for English typing and syntax errors.

Comments from Reviewer Malgorzata

Major revisions
1: Results of biochemical data has been included in table 1 and mentioned in the result section.
2: Results of frequency & distribution of CYP21A2 gene mutations has been presented as graph (figure 2)
3: Spectrum and frequency of CYP21A2 gene mutations has been compared with other populations in table 2

Minor revisions
1: It is not possible to increase the sample size at this stage. However, the purpose of this study is to provide a service where the testing for CAH diagnosis can be made available in the country. We hope to do this in future.
2: It is not possible to screen for more mutations at this stage.

I hope that the answers to the reviewer’s queries are satisfactory.

Regards,
Dr Tariq Moatter