Reviewer's report

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

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Reviewer: Malgorzata M. Wasniewska

Reviewer's report:

This is a simple report of the mutation spectrum of CYP21A2 gene in the polyethnic Pakistani population. Authors presented a cross section study of 29 patients with classical forms (SW and SV) of CAH. This is an experience of one Endocrinological Center.

With the knowledge of the distribution of 8 more frequent CYP21A2 gene mutations, the Authors would like to promote the CAH prenatal diagnosis program in the local population, because only the 41% of the classical CAH cases of that study population were diagnosed during the newborn period. Moreover Authors suggested that with high degree of parental consanguinity (65.5%) in that study population, there is the need of medical facilities with genetic testing and counseling for CAH patients to help consanguineous couples to make informed and intelligent reproductive decisions. Authors also underlined that so far the cultural norms with a long tradition of making family-centered health care decisions take to failure the diagnosis and treatment of CAH in that population.

Considering the cultural and ethnic diversity, that study (may have) can play a social role to promote the neonatal CAH screening program in that country.

Major Compulsory Revisions:

1. Lack of biochemical (hormonal) results, for CAH diagnosis, of study population; may be useful to show at least 17OHP results.

2. It can be useful to present the results of frequency and distribution of CYP21A2 gene mutations as graphs.

3. It can be also interesting to discuss the spectrum and frequency of CYP21A2 gene mutations in Pakistani population and compare it with other populations just reported in the literature to underline the eventual peculiarity of molecular picture in that study group.

Minor Essential Revisions:

1. Small study group to be representative for the nation.

2. The samples of DNA were screened for only 8 more frequent mutations of CYP21A2 gene.

Level of interest: An article of limited interest
Quality of written English: Acceptable

Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:

I declare that I have no competing interests' below.