Reviewer's report

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

**Version:** 2  **Date:** 25 October 2010

**Reviewer:** Leonidas Phylactou

**Reviewer's report:**

This report by Khan and colleagues represents a description of the prevalence of eight common mutations of the 21-hydroxylase gene in CAH patients.

This is an interesting report which provides evidence about the incidence of CAH mutations in Pakistan.

**Major compulsory revisions**

1. As it stands, the report contains preliminary results. In certain cases the authors report that PCR products were not generated probably due to the presence of deletions. Furthermore, the presence of multiple mutations in single patients poses the question whether this is due consanguinity or major DNA rearrangement or deletion. It would be appropriate to perform large fragment detections (e.g. by MLPA, real-time PCR or Southern blotting) to determine such presence in the samples. Moreover, in the cases of multiple mutations it is important to perform genetic analysis on the parents of the affected individuals.

2. The discussion is very long compared to the rest of the document and should be restructured in order to be more balanced.

3. A figure with the amplified fragments, targeted mutations and primers would be useful.

**Minor essential revisions**

1. In the results section, fourth line, the female to male ration should be 15:14 (i.e. 1.07).

2. In the introduction, it should be specified whether the patients used in this study were included in previous published papers of the same group.

3. The manuscript should be corrected for English typing and syntax errors.

**Level of interest:** An article whose findings are important to those with closely related research interests

**Quality of written English:** Needs some language corrections before being published

**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