Reviewer's report

Title: Novel Deletion Alleles Carrying CYP21A1P/A2 Chimeric Genes In Brazilian Patients With 21-Hydroxylase Deficiency

Version: 1 Date: 31 December 2009

Reviewer: SABINA BAUMGARTNER-PARZER

Reviewer's report:

The experimental work performed in the present study is sound and acknowledged. The authors should clarify to which group of patients or population the "9%" of chimeric A2A1P genes refer.

The authors give ratios of the different probe-signals in mlpa (2:1 or 3:2) and it is not clear to this reviewer, how the cofidence intervals for the different mlpa-probes have been determined.

What is the meaning of instead in the sentence "...novel and rare mutations, respectively, instead of.." (abstract, results).

Referring to and addressing founder effects, HLA-haplotypes would have been of interest in the carriers of deletions and chimeric genes. In general, the authors should try to shorten the different sections, particularly the "background", and to focus the discussion. The impact of the study for research as well as potential implications for the patient and the respective therapy should be addressed and specified. It would be helpful for reading of the "Results" section that the latter could be more structured - e.g - according to patients' numbers or other criteria.

What do the authors suggest concerning the methods applied: mlpa and southern blotting in all cases, or is mlpa sufficient in addition to sequence analysis?

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

Quality of written English: Not suitable for publication unless extensively edited

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.