Reviewer's report

Title: Novel p.Cys65Tyr mutation in NR5A1 gene in three 46,XY siblings with normal testosterone levels and signs of late adrenal insufficiency and their mother with primary ovarian insufficiency

Version: 2 Date: 25 October 2013

Reviewer: John Achermann

Reviewer's report:

The authors have addressed most of the points raised but I still have some issues related to these

Minor Essentials Revisions

1) It is excellent that low dose synacthen tests have been performed and earlier data are available for ACTH levels. These data are very important but have weakened the argument for adrenal insufficiency. Even the proband has a response that is just suboptimal, and the ACTH values that are raised are only just above normal limits. I do agree that long term monitoring is needed but the constant referrals to adrenal insufficiency in the title, abstract, end of introduction and in the case report is overplayed. At the very least "possible" adrenal insufficiency should be used though I would be very cautious here.

2) Abstract: the apparent normal Leydig and Sertoli cell function is ON POSTNATAL TESTING. There was clearly an issue during development.

3) The written English has got worse in places. Many articles ("the" "a") are missing and sentences have been edited to make them worse (eg "The SF-1 protein has 461 amino acid" should have been modified to "amino acids" rather than "SF-1 protein has 461 amino acid"). The case report changes need much attention. Hormone "dosages" should be measurements/concentrations or levels.

Level of interest: An article of importance in its field

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