Author's response to reviews

Title: The novel p.Cys65Tyr mutation in NR5A1 gene in three 46,XY siblings with normal testosterone levels and their mother with primary ovarian insufficiency

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To
BMC Medical Genetics

Dear Sirs,

This is to submit the revised version of manuscript now entitled “The novel p.Cys65Tyr mutation in NR5A1 gene in three 46,XY siblings with normal testosterone levels and their mother with primary ovarian insufficiency” to be considered for publication in BMC Medical Genetics as a Case Report.

We have addressed all comments and criticisms received from referee to whom we present our appreciation. Details are presented below.

We thank you for your attention.

Yours sincerely,

Maricilda Palandi de Mello, Ph.D.

Answers to Reviewer’s report

Reviewer: John Achermann

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

Minor Essential 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.

We corrected the title and the manuscript giving less emphasis on the adrenal insufficiency.

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

Yes.

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.

We have revised English throughout the manuscript.