Reviewer’s report

Title: The novel p.Cys65Tyr mutation in NR5A1 gene in three siblings with 46,XY disorders of sex development

Version: 1 Date: 16 August 2013

Reviewer: Giovanni Neri

Reviewer’s report:

Major compulsory revision:
The authors should report more clearly on the gonadal status, especially of the older sib. Were the testes developing normally? What is the prognosis? Will these individuals be fertile or not?

Minor revision:
The report is generally well balanced, perhaps with too much emphasis on the attempt at convincing the readers that the described mutation is truly pathogenic. A more thorough description of the maternal phenotype would be helpful. Why was she considered to be on her way to primary ovarian insufficiency? The authors should discuss the possibility that she is essentially not affected because the effect of the mutation is limited to the male sex. In fact, it would be good to know if there are in the mother’s family other individuals that could be tested as possible carriers of the mutation. If this were a numerous family, the information to be gained from extended gene testing would be significant.

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:

No competing interest