Reviewer's report

Title: Adult phenotype and further phenotypic variability in SRD5A3-CDG

Version: 2

Date: 30 November 2013

Reviewer: Fernando Scaglia

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Major Compulsory Revisions

The authors should create a table so the molecular, biochemical and clinical features of these two siblings could be compared with other patients with SRD5A3-CDG and with Kahrizi syndrome patients. A table would help to better delineate the broad phenotypic spectrum associated with this type of CDG.

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

Quality of written English: Acceptable

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.