Author's response to reviews

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

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Professor Dr. Jörg T. Epplen

Section Editor, BMC Medical Genetics

Dear Professor Epplen:

Thank you for having our manuscript reviewed. We amended the manuscript according to the comments of the reviewers.

We think that the manuscript has improved considerably and hope that it now meets the standards of the journal.

Sincerely,
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Response to Reviewers

We thank reviewers Dr. Lynne A Wolfe and Dr. Fernando Scaglia for evaluating our manuscript and for the useful comments. We prepared a table in accordance with the suggestions of both reviewers and added to the end of the manuscript.

We checked PubMed to make sure that no new cases were reported.

Reviewer I

Question 1. *Mild intellectual disability seems inappropriate for a 40 yr old described as requiring assistance for ADL's. Was formal neuropsych testing accomplished?*

**Response:** Formal neuropsychological testing could not be performed, because the parents of the patients did not agree for the test. The parents did not agree to cranial magnetic resonance imaging, either.

Question 2. *Skeletal survey content would be important information since subtle bone abnormalities has been reported in other patients with SRD5A3-CDG. A table comparing these sibs to other SRD5A3-CDG as well as Kahrizi cohort would improve ease of understanding the phenotype.*

**Response:** We agree that skeletal survey would have been useful, but the parents did not agree to it. A table comparing our patients to other patients reported with *SRD5A3* mutation is added. Please see table 1 at the end of the manuscript.
Reviewer II

*Question 1.* 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 better delineate the broad phenotypic spectrum associated with this type of CDG.

**Response:** A table comparing our patients to other patients reported with *SRD5A3* mutation is added. Please see table 1 at the end of the manuscript.