Author's response to reviews

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

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Author's response to reviews: see over
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Professor Dr. Jörg T. Epplen
Section Editor, BMC Medical Genetics

Dear Professor Epplen:

Thank you for the three minor comments. We amended the manuscript accordingly and hope that it now meets the standards of the journal.

Best Wishes for the New Year.

Sincerely,
Aslı Tolun, PhD

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Response to Minor Amendments Requested:

1. The authors state that their 40 year-old patient has mild intellectual disability. However, he requires assistance for ADLs (activities of daily living). That is not consistent and they need to consider changing that statement to moderate or severe intellectual disability.

We could not perform mental performance tests for either patient. Thus, mental status was evaluated only by the observations of the clinician. The older patient requires assistance for activities of daily living, but the main reason for it is visual problems rather than intellectual disability. Nonetheless, we agree that it will be more consistent to change “mild intellectual disability” to “moderate intellectual disability” for the older patient, as requested.

2. The other patient who is the sibling of this 40 year old is described as being married and having "four healthy siblings". I think the authors probably wanted to state that he had "four healthy children or offspring" rather than siblings. They need to change this sentence accordingly.

We changed the statement “four healthy siblings” to “four healthy children”.

3. This younger sibling was described as able to finish elementary or primary school but not able to finish high school. I would also argue that this individual has intellectual disability. Thus, in the Table the authors should state that both patients have intellectual disability and not one."

Our observations for his activities of daily living and at work were consistent with borderline mental status. We think that he could not continue his education beyond primary school mainly because of visual problems. We amended Table 1 as both patients having intellectual disability, as suggested.

We emphasized visual problems during the patients’ education.

We thank for the useful comments.