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:

We are submitting our manuscript entitled “Adult phenotype and further phenotypic variability in SRD5A3-CDG” for consideration for publication as a case report. The manuscript describes the clinical phenotypes of two brothers aged 38 and 40 years; all patients reported previously are children. Our patients have unusually late onset and mild clinical phenotypes than all patients with SRD5A3-CDG reported so far. They also have atypical ocular findings and variable phenotypes. We discovered a homozygous truncation mutation in \textit{SRD5A3} with a homozygous nonsense mutation. The mutation had been reported previously.

All authors have been involved in drafting the article and revising it critically, and read and approved the submission to BMC Medical Genetics. No part of the work has been published previously in print or electronic format, the manuscript is not under consideration for publication elsewhere, and we do not have any related manuscripts in preparation or submitted. The authors have no conflict of interest to declare. Please find below the names and contact information for two suggested reviewers.

Sincerely,

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