Author’s response to reviews

Title: Familial Xp11.22 microdeletion including SHROOM4 and CLCN5 is associated with intellectual disability, short stature, microcephaly and Dent disease

Authors:
Magdalena Danyel (magdalena.danyel@charite.de)
Eun Kyung Suk (eun-kyung.suk@humangenetik-berlin.de)
Vera Raile (vera.raile@charite.de)
Jutta Gellermann (jutta.gellermann@charite.de)
Alexej Knaus (alexej.knaus@ukbonn.de)
Denise Horn (denise.horn@charite.de)

Version: 2 Date: 26 Dec 2018

Author’s response to reviews:

MGNM-D-18-00275R1
Familial Xp11.22 microdeletion including SHROOM4 and CLCN5 is associated with intellectual disability, short stature, microcephaly and Dent disease Magdalena Danyel; Eun Kyung Suk; Vera Raile; Jutta Gellermann; Alexej Knaus; Denise Horn BMC Medical Genomics

Dear Editor Dr. Menard,

thank you for your comments. I made all changes. For my answers: Please, see below.
Best wishes, Denise Horn

Dear Prof. Horn,

Your manuscript "Familial Xp11.22 microdeletion including SHROOM4 and CLCN5 is associated with intellectual disability, short stature, microcephaly and Dent disease" (MGNM-D-18-00275R1) has been assessed by our reviewers. Based on these reports, and my own assessment as Editor, I am pleased to inform you that it is potentially acceptable for publication in BMC Medical Genomics, once you have carried out some essential revisions:
1. Please specify in the Consent for publication section who consented on behalf of the patient, who is a minor. Please confirm if this consent was written or verbal. If verbal, please state the reason and whether the ethics committee approved this procedure.

I got a written consent from the parents.

2. As it is clearly stated in the attached consent form that only "anonymised" images of the patient can be published, please anonymise the image of the patient. We would suggest a back box covering the patient's eyes.

For us, “anonymised” means a clinical description without calling the name of the patient. But I see that the sentence is unclear therefore I covered the eyes of the patient with a black box.

3. Please remove the signed consent form at this stage.
I did it.

4. Please ensure the figures, especially figure 2, are of high quality.
I improved the quality of figure 2 and changed figure 1.

5. Please amend the main text headings to: Background, Case Presentation, Discussion & Conclusions. There are currently many extra headings, such as Methods, which are not standard.
I removed the headings.

6. At this stage, please upload your manuscript as a single, final, clean version that does not contain any tracked changes, comments, highlights, strikethroughs or text in different colours. All relevant tables/figures/additional files should also be clean versions. Figures (and additional files) should remain uploaded as separate files.
I made these changes.

Once you have made the necessary corrections, please submit a revised manuscript online at:
https://mgnm.editorialmanager.com/

If you have forgotten your password, please use the 'Send Login Details' link on the login page at https://mgnm.editorialmanager.com/. For security reasons, your password will be reset.

Please also ensure that your revised manuscript conforms to the journal style, which can be found at the Submission Guidelines on the journal homepage.

A decision will be made once we have received your revised manuscript, which we expect by 26 Dec 2018.

Please note that you will not be able to add, remove, or change the order of authors once the editor has accepted your manuscript for publication. Any proposed changes to the authorship must be requested
during peer-review, and adhere to our criteria for authorship as outlined in BioMed Central's policies. To request a change in authorship, please download the 'Request for change in authorship form' which can be found here - http://www.biomedcentral.com/about/editorialpolicies#authorship. Please note that incomplete forms will be rejected. Your request will be taken into consideration by the editor, and you will be advised whether any changes will be permitted. Please be aware that we may investigate, or ask your institute to investigate, any unauthorized attempts to change authorship or discrepancies in authorship between the submitted and revised versions of your manuscript.

We look forward to receiving your revised manuscript and please do not hesitate to contact us if you have any questions.

Best wishes,

Anne Menard
BMC Medical Genomics
https://bmcmedgenomics.biomedcentral.com/