Reviewer’s report

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

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Reviewer: Felix Claverie-Martin

Reviewer's report:

It was a pleasure to review the manuscript "Familial Xp11.22 microdeletion including SHROOM4 and CLCN5 is associated with intellectual disability, short stature, microcephaly and Dent disease: A Case Report" by Danyel and col. submitted to BMC Medical Genomics. The authors present the case of a 4-year-old boy with short stature, speech delay, mild intellectual disability, microcephaly, and facial dysmorphism, as well as the clinical characteristics of a rare renal tubulopathy known as Dent disease. By karyotyping analysis, they found a microdeletion Xp11.23p.11.22 that was inherited from the patient's mother. This microdeletion includes the SHROOM4 and CLCN5 genes. SHROOM4 has been shown to be associated with X-linked intellectual disability. On the other hand, the deletion of CLCN5 explains the renal tubulopathy in the patient. Only one previous similar case with Xp11.22 microdeletions that include CLCN5 and SHROOM4 has been reported. The manuscript is well written and the results are clear. I only have a minor comment:

The authors indicate that the patient's mother is asymptomatic. With regard to Dent disease this is normal in the patients' mothers (the patients are usually males), but how do you explain the absence of developmental delay in the mother of this patient?

Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

Yes

Does the work include the necessary controls?
If not, please specify which controls are required in your comments to the authors.

Yes

Are the conclusions drawn adequately supported by the data shown?
If not, please explain in your comments to the authors.

Yes
Are you able to assess any statistics in the manuscript or would you recommend an additional statistical review?
If an additional statistical review is recommended, please specify what aspects require further assessment in your comments to the editors.

Not relevant to this manuscript

Quality of written English
Please indicate the quality of language in the manuscript:

Acceptable

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