Reviewer’s report

**Title:** EXTENDED CLINICAL FEATURES ASSOCIATED WITH NOVEL GLIS3 MUTATION: A CASE REPORT

**Version:** 0  **Date:** 28 Nov 2016

**Reviewer:** John Gregory

**Reviewer's report:**

This case report describes an infant from a highly consanguineous background with a mutation of the GLIS3 gene and a phenotype that extends the range of abnormalities reported in the literature to include a range of genital abnormalities including hypospadias, chordee and bifid scrotum. Unfortunately, I don't think this case report is publishable as it is unclear whether this mutation is responsible for the range of genital abnormalities reported in this patient. Such genital abnormalities are relatively common and even though there is no underlying explanation given the apparently normal endocrine responses to HCG stimulation, the authors do not provide any evidence that these abnormalities are a consequence of the GLIS3 mutation rather than a coincidental finding or a consequence of another genetic abnormality arising from the underlying consanguinity.

**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.

No

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

No

**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:
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