Reviewer’s report

Title: Case report of whole genome sequencing in the XY Female: identification of a novel SRY mutation and revision of a misdiagnosis of androgen insensitivity syndrome

Version: 0  Date: 06 Oct 2016

Reviewer: Thomas F. Kolon

Reviewer's report:

This is a very good review of CAIS vs XY CGD.

The authors should comment that the simple pathologic demonstration of bilateral streak gonads during laparoscopy at age 18 yrs should have prompted immediate further genetic evaluation rather than 26 yrs later. Was there any evidence of microscopic gonadoblastoma or dysgerminoma in the streak gonads given the presence of a Y chromosome?

The Background section is very comprehensive and the appropriate description of the differences between CAIS and CGD might be better served in a Discussion after the Case Presentation.

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.

Unable to assess

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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