Reviewer’s report

Title: 48,XXYY syndrome presenting with long-term infertility and newly observed neck deformities.

Version: 0 Date: 24 Jan 2020

Reviewer: Adriano Alberti

Reviewer's report:

1. Do you believe the case report is authentic?
   Yes.

2. Do you have any ethical concerns? Please consider if local Institutional Review Board approval or ethical approval was obtained (if appropriate) and if the patient (or their parent or guardian in the case of children under 18) gave written, informed consent to publish this case and any accompanying images. A statement to this effect should appear in the manuscript.
   Comments: Yes, the ethical question of the study is appropriate.

3. Does the Introduction explain the relevance of the case to the medical literature?
   Yes.

4. Does the article report the following information? Where information is missing, please specify.
   a. The relevant patient information, including:
      - De-identified demographic information (age, gender, ethnicity)
      - Main symptoms of the patient
      - Medical, family and psychosocial history
      - Relevant past interventions and their outcomes
   b. The relevant physical examination findings
   c. Important dates and times in this case (if appropriate, organized as a timeline via a figure or table); if specific dates could lead to patient identification, consider including time relevant to initial presentation, i.e. initial presentation at T = 0, follow up at T = 1 month.
   d. Diagnostic assessments, including:
      - Diagnostic methods
      - Challenges (e.g., financial, language/cultural)
      - Reasoning and prognostic characteristics (e.g., staging), where applicable
   e. Types and mechanism of intervention
   f. A summary of the clinical course of all follow-up visits
   Comments: Yes, this is adequately presented.
5. Is the interpretation (discussion and conclusion) well balanced and supported by the case presented? Comments: Yes.

6. Is the anonymity of the patient protected? Please consider any identifying information in images such as facial features or nametags, whether the patient is named etc. If not, please detail below. Yes.


8. Does the case represent a useful contribution to the medical literature? Comments: Yes.

9. Additional comments for the author(s)? Comments: Thank you for the opportunity to review this report. This report can be improved for possible publication, eady the recommendations below:

Include a description of the patient's relevant demographic details.
Insert more references in the discussion

**Level of interest**
Please indicate how interesting you found the manuscript:

An article of importance in its field

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

Acceptable

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Please complete a declaration of competing interests, considering the following questions:

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No