Reviewer’s report

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

Version: 0 Date: 26 Jan 2020

Reviewer: Walid AL ACHKAR

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: no

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) y
      - Main symptoms of the patient y
      - Medical, family and psychosocial history y
      - Relevant past interventions and their outcomes y
   b. The relevant physical examination findings y
   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. y
   d. Diagnostic assessments, including:
      - Diagnostic methods y
      - Challenges (e.g., financial, language/cultural) y
      - Reasoning and prognostic characteristics (e.g., staging), where applicable y
   e. Types and mechanism of intervention y
   f. A summary of the clinical course of all follow-up visits y

Comments:
a rare case with progressive testicular failure accompanied with
48,XXYY syndrome

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

7. Is the Abstract representative of the case presented?
   Comments: yes

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

9. Additional comments for the author(s)?
   fig 3 to 5 need more of your attention put on fig 3 each line name , on FISH picture you have to put the probe name and manufacture company

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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No