Author’s response to reviews

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

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Author’s response to reviews:

Reviewer #1: Thank you for the decision of acceptance, this case shows how underlying genetic factors for many cases of infertility can go undetected for years especially when coupled with an atypical presentation.

Reviewer #2: Thank you for your valuable contribution.
About the final comments: 1. Marital status was added in page 4 line 54 and highlighted in red but additional demographic details would not comply with the journal's policy of anonymization.
2. Additional recent references were added to the discussion.

Reviewer #3: Thank you for your valuable contribution.
About the final comments: 1. About figure 3: The interpretation system used for the test results for the A,B,C,D and E segments of the figure along with the manufacturing company were added in page 5 line 77 and highlighted in red.
2. About figure 5: The FISH probe name and manufacturing company info were added in page 6 line 87 and also highlighted in red with colors used for the test result in the figure also added in the figure caption.

Reviewer #4: Thank you for your valuable contribution.
Question 4 Section b: The highlight of this case is the atypical presentation itself spanning years undetected including many atypical features such as short stature.

Reviewer #5: Thank you for your valuable contribution.
Question 3: Delays in development was corrected to delayed development in page 3 line 45 and highlighted in red. Said abnormalities was also reworded in page 3 line 46 and also highlighted in red.
Question 4 Section b: No characteristic facial features were noted although the patient's face with eyes hidden behind a black box was submitted to the journal along with Figure 1 but the figure had to be cropped as supplementing the patient's face does not comply with journal policy. The patient's span wasn't noted to be abnormal prior to genetic diagnosis but the patient had an atypical short stature.
Section d: Hypergonadotropic hypogonadism shown in page 5 line 70 was the major constituent of the reasoning written in page 5 line 81 and a suggestive feature for workup towards identifying the cause behind testicular failure.
No followup visits were mentioned as the patient was only interested in diagnosis of the cause behind his infertility and thus was uninterested in any followup visits that wouldn't contribute to conception after diagnosis.
The mentioned beta-hCG testing and its result was added from our patient info to the manuscript in
page 5 line 71 and highlighted in red.
Section f: Again the patient was uninterested in any followup visits.
Question 5: The conclusion was reworded to better show the case conclusion and be more conclusive.
Final comments: Syntax and words pointed out were corrected and highlighted in red. The correct version of the abstract will be resubmitted in the revised version of the manuscript.