Author’s response to reviews

Title: Premature ovarian insufficiency as a variable feature of blepharophimosis, ptosis, and epicanthus inversus syndrome associated with p.Leu75Phe FOXL2 mutation: a case report.

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Author’s response to reviews:

We enclose a revised version of the paper entitled “Premature ovarian insufficiency as a variable feature of blepharophimosis, ptosis, and epicanthus inversus syndrome associated with p.(Leu75Phe) FOXL2 mutation: a case report”, which was previously submitted for publication in the BMC Medical Genetics (MGTC-D-18-00417).

We are very grateful for giving us the opportunity to revise the manuscript. We would also like to thank all the Reviewers for their time and all the valuable remarks, which allowed us to improve our paper. We tried to answer all the questions and correct whatever was suggested. All the changes in the manuscript are marked in yellow.

Point-by-point response to all the remarks:

Responses for the Reviewers:

Ad. Reviewer 1 (Petra Liskova):

‘Abstract should indicate whether the mutation is novel.’ - this information has been already introduced to the abstract session.

‘ The mutation in the abstract should be also described at DNA level not only at the protein level.’ - this information has been already introduced to the abstract session.
"The following case study is the first report of endocrine impairments typical for menopausal transition in young female with FOXL2 gene mutation previously associated with BPES type 2.' – this sentence has been already clarified. The authors mean that only p.(Leu75Phe) FOXL2 mutation has not been associated with different phenotypes yet.

The introduction session has been already reworded.

Minor comments:

1. Revised.
2. Revised.
3. Revised.
4. Revised.
5. Revised.
6. Revised.
7. Revised.
8. Additional reference was added.
9. Revised.
10. This information was moved to the discussion section.
11. Revised.
12. Revised.
13. Revised.
15. Revised.
16. Revised
17. Revised
18. According to the ‘instructions for the authors’, each figure should be uploaded separately.
19. Revised
20. Revised

Ad. Reviewer 2 (Yueqiu Tan):

BPES types has been corrected throughout the text.

All authors have read the revised version of the manuscript and approved its contents.

Thank you very much for considering our manuscript for publication.

Yours sincerely,

Damian Warzecha MD,

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