Reviewer’s report

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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Reviewer: Jeehyeon Bae

Reviewer's report:

This manuscript entitled "Premature ovarian insufficiency as a variable feature of blepharophimosis, ptosis, and epicanthus inversus syndrome associated with p.Leu75Phe FOXL2 mutation: a case report." by Grzechocińska et al is reporting a case of the BPES patient possessing the p.Leu75Phe FOXL2 mutation. Manuscript has concisely written and is informative. Authors performed whole exome sequencing of the proband of suspected BPES and identified p.Leu75Phe mutation in FOXL2 without plausible rare variants in other genes. Although this particular mutation type is not newly identified in this study, hormonal profiles provided are exceptionally useful information for those whom study BPES and/or POI. I feel that this manuscript is acceptable for the publication in BMC Medical Genetics.

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?
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Yes

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Yes

Are you able to assess any statistics in the manuscript or would you recommend an additional statistical review?
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Not relevant to this manuscript
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Acceptable

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