Reviewer’s report

Title: Novel FOXL2 mutations in two Chinese families with blepharophimosis-ptosis-epicanthus inversus syndrome

Version: 1 Date: 2 June 2015

Reviewer: Hannah Verdin

Reviewer’s report:

Major Compulsory Revisions

The discussion is rather short to my opinion. The authors should elaborate on the deletion of the polyalanine tract as this has never been described in a BPES patient to my knowledge but it has been described in a patient with POF (Harris et al. 2002). The different studies describing the functional consequences of the polyalanine tract should also be discussed in more detail and the study by Moumné et al. (Human Molecular Genetics 2005) should be added to this discussion as they investigate the functional consequences of a deletion of the polyalanine tract.

Also, the discussion on the missense mutation could be more extensive. For example, discussing the molecular consequences, an overview of all missense mutations in the FOXL2 forkhead domain, genotype-phenotype correlation, etc.

Minor Essential Revisions

General remarks:

1. Use italic when the gene is meant and plain (roman) for when the protein is meant.
2. Check the use of a space after round brackets, commas, …
3. The full name of FOXL2 is forkhead box L2 and not forkhead transcription factor 2, please correct this.
4. HGVS recommends the use of the three-letter abbreviations for amino acids in mutation descriptions on protein level, please change p.L75F to p.Leu75Phe.

Specific remarks:

5. Do all the affected family members of both Chinese families have the mutation? This is not clear from the article and indicating the genotype of the screened family members (normal and affected) on the pedigree would clarify this.

6. What was the result from screening the 100 controls?

Typos and word use:

7. Page 2, line 42: delete “the” before Chinese families (also on page 4, line 67)
8. Page 3, line 53 and 54: replace ocular with eyelid
9. Page 3, line 63: correct granulose to granulosa
10. Page 3, line 65: insert a space between in and regulating
11. Page 6, line 120: intellectual disability is preferred instead of mental retardation
12. Page 6, line 129: delete gene after FOXL2
13. Page 6, line 132: replace the last comma by “and”
14. Page 7, line 136: delete putative
15. Page 7, line 145: delete “the” before our study
16. Page 8, line 163: change poly-phenylalanine tract to polyalanine tract
17. Page 9, line 177: change poly-phenylalanine tract to polyalanine tract

**Level of interest:** An article whose findings are important to those with closely related research interests

**Quality of written English:** Needs some language corrections before being published

**Statistical review:** No, the manuscript does not need to be seen by a statistician.

**Declaration of competing interests:**

I declare that I have no competing interests