Reviewer’s report

Title: Targeted next-generation sequencing in anophthalmia and microphthalmia patients confirms SOX2, OTX2 and FOXE3 mutations

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Reviewer: Fernando Scaglia

Reviewer’s report:

The manuscript submitted by Lopez-Jimenez and collaborators entitled "Targeted Next-Generation Sequencing in Anophthalmia and Microphthalmia patients confirms SOX2, OTX2 and FOXE3 mutations" is an article of importance and merit in its field as it clearly shows the power of next-generation sequencing for the rapid screening of candidate genes for the specific birth defects of microphthalmia and anophthalmia. The authors were able to identify disease-causing mutations and novel sequence variants. Nevertheless, next-generation sequencing was not found to be as useful to detect small intragenic deletions and duplications. Mutations were not found in 10 of the patients which led the authors to conclude that there is genetic heterogeneity in anophthalmia and microphthalmia. The results of the study also emphasized the rarity of mutations in some of the known pathogenic genes and the need for finding other genes connected with these two conditions.

The questions posed by the authors were well defined and the methods they used for the study were appropriate and well described. The data were sound and I consider that the manuscript adheres to the relevant standards for reporting and data deposition. Overall the discussion and conclusions were written in a balanced manner and are adequately supported by the data.

Level of interest: An article of importance in its field

Quality of written English: Acceptable

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.