Reviewer's report

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

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Reviewer: Momiao Xiong

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This paper attempts to demonstrate the power of next-generation sequencing with pooled sample groups for the rapid screening of candidate genes for birth defects as we were correctly able to identify disease-causing mutations and novel sequence variants. They also found that next-generation sequencing was less useful for small, intragenic deletions and duplications. These results may be useful for clinical diagnosis. However, due to the small sample size, it is unclear that what false positive and negative rates to identify mutations using next-generation sequencing. Since the coverage in the experiments is very high it is also useful to report the results when the coverage is reduced.

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

Quality of written English: Acceptable

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

Declaration of competing interests:

No