Reviewer’s report

Title: CYP1B1 mutation studies in patients with primary congenital glaucoma from Saudi Arabia, and identify patients with possible novel mutations in new glaucoma genes

Version: 1 Date: 20 May 2014

Reviewer: Nobuo Fuse

Reviewer’s report:

Primary congenital glaucoma is a relatively rare cause of glaucoma typically inherited as an autosomal recessive manner with variable penetrance with prevalence of childhood blindness ranges from 0.03% in developed countries to 0.12% in undeveloped countries.

The authors provided a mutation spectrum in Saudi PCG patients. This article is intriguing for the physician.

I have some point to need to be addressed.

Minor Essential Revisions

Major points

1. The authors mentioned that “This study was conducted to identify CYP1B1 mutations in Saudi PCG patients with various ethnic backgrounds, and to identify those patients who carry mutations in novel genes.” However they screened the only CYP1B1 gene. So it was overstated. (lines 70-71)

2. Total six patients showed heterozygous mutation. Was there no compound heterozygotes? Please mention about it.

3. In Table 1, Mean C/D was different between Native Saudi and Non-native Saudi. It is better that the information of surgical intervention would be included.

Minor point

Page 7 line 124, refraction would be -4.92.

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:

There is no competing interests in relation to the paper they are reviewing.