Reviewer’s report

Title: Progressive Optic Nerve Changes in Cavitary Optic Disc Anomaly: integration of copy number alteration and cis-expression quantitative trait loci to assess disease etiology

Version: 0 Date: 18 Dec 2018

Reviewer: Takeshi Iwata

Reviewer's report:

The manuscript "Progressive Optic Nerve Changes in Cavitary Optic Disc Anomaly: integration of copy number alteration and cis-expression quantitative trait loci to assess disease etiology" by Hwang et al., describes 4 affected and 14 unaffected single pedigree with CODA. Using the long-range PCR, authors determines the precise size and tandemly located CNA on chromosome 12. Six SNPs within CAN region were identified, which acts as eQTLs for multiple genes.

This manuscript adds novel information about CODA, another optic nerve related disease by copy number alteration (CNA). Authors have characterized CNA at accuracy never done before. It would be nice if authors can demonstrate gene expression changes in brain and nerve related genes in Figure 5.

Minor correction:
Page 9, line 11-13 - 1.1 bp product
Page 13, line 8, p published

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?
If not, please specify which controls are required in your comments to the authors.

Yes

Are the conclusions drawn adequately supported by the data shown?
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Yes
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I am able to assess the statistics

**Quality of written English**
Please indicate the quality of language in the manuscript:

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