Reviewer’s report

Title: A Method for Determining Haploid and Triploid Genotypes and their Association with Vascular Phenotypes in Williams Syndrome and 7q11.23 Duplication Syndrome

Version: 0 Date: 14 Jan 2018

Reviewer: Susan Corley

Reviewer's report:

The main purpose of this paper is the reporting of a method for identifying haploid and triploid genotypes in CNV regions. Williams syndrome patients (WS) and Dup7 patients are tested for proof of concept. Furthermore genotype-phenotype associations are made in the context of aortic pathology in WS and Dup7.

The choice of Williams syndrome and Dup7 for testing is clever and allows interesting genotype-phenotype comparisons to be made.

My concern is that the methodological advances are not clearly described in this paper. The authors say they have used PennCNV software and then internally generated R scripts. The algorithms in the R scripts are not described. Similarly there is not a clear description of how "Tests of the association of the degree of SVAS with every SNP in 7q11.23 .." (p6 line 15) were performed.

The short description of methods provided in the paper under review might be contrasted with that by Lin et al. Analyzing Copy Number Variation using SNP Array Data Protocols for Calling CNV and Association Tests 2014 Curr Protoc Hum Genet. It is not clear whether the authors of the paper under review have deviated significantly from the approaches described by Lin et al. and if so how.

As a minor point the Figures are not well resolved, I expect higher quality images will be required.

Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

Unable to assess

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

Unable to assess
Are the conclusions drawn adequately supported by the data shown?
If not, please explain in your comments to the authors.

Yes

Are you able to assess any statistics in the manuscript or would you recommend an additional statistical review?
If an additional statistical review is recommended, please specify what aspects require further assessment in your comments to the editors.

I recommend additional statistical review

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

Acceptable

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