Author's response to reviews

Title: Detecting 22q11.2 deletion in Chinese children with conotruncal heart defects and single nucleotide polymorphisms in the haploid TBX1 locus

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Version: 4 Date: 12 November 2011

Author's response to reviews: see over
Dear Editor,

We appreciate all of the enlightening and helpful comments from the reviewers. The manuscript has been revised accordingly, and the modified parts are highlighted in blue. We have also responded to each reviewer’s comments point-by-point as listed below.

Thank you very much for your consideration of our manuscript.

Best regards

Yours sincerely,

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Response to reviewer's comments:

Reviewer: Dana Crawford
Comments to the Author
Minor essential revisions:

Author's response:

Thank you for pointing this out. We have performed tests of Hardy Weinberg Equilibrium only for non-deletion patients and healthy controls. We have made changes accordingly in the revised manuscript. Please see the subsection titled “Gene PCR and sequencing” in the Result section.

Other final, minor points:
1. In Table 3, the authors should replace “dbSNP” with “HapMap”. And, they should indicate in the table or legend which HapMap population is being represented. dbSNP actually contains a lot more data than just HapMap, so the current label is vague and confusing. Likewise, the HapMap website and population should be mentioned either in the Methods section or in the Results section (under Gene PCR and sequencing, which mentions dbSNP).

Author's response:
We downloaded the allele_freqs_chr22_CHB_phase3.2_nr.b36 from HapMap website.
However, it only has three SNPs (i.e. rs2301558, rs5746826 and rs4819522), and lacks the other five of the eight SNPs discussed in our manuscript in the database. Then we looked up them in the 1000 Genome Project Database, and used the allelic counts and individual sample data of the eight SNPs in this Database. We have made changes accordingly in the revised manuscript. Please see the subsection titled “Gene PCR and sequencing” in the Result section and Table 3.

2. The authors state in their response to previous reviews that “due to lack of the accurate number of the dbSNP cohort, we could not perform comparisons between dbSNP and our cohort.” This is an odd statement given that HapMap data (not dbSNP, as mentioned above) can be downloaded for analysis. Therefore, the allelic and genotypic counts and individual sample data are available for analysis presented in Table 4.

Author’s response:
We have made changes accordingly in the revised manuscript. The allelic and genotypic counts and individual sample data came from the 1000 Genome Project Database. Please see the Table 4.