Reviewer’s report

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

Version: 3 Date: 23 September 2011

Reviewer: Dana Crawford

Reviewer’s report:

Minor essential revisions:

The authors have addressed most of the concerns by this Reviewer. There is one additional point that this Reviewer was remiss in including in previous reviews. That is, the authors calculated Hardy Weinberg Equilibrium for the variants identified among 22q deletion patients (or, at least that what is seems according to the statement “All eight SNPs of HWE_P values were >0.05” in the subsection titled “Gene PCR and sequencing”). This does not make sense given these patients are haploid at these variants. These patients only have one allele or the other; there are no heterozygotes or homozygotes. Tests of Hardy Weinberg Equilibrium are performed to determine if there is an excess of homozygosity or heterozygosity due to poor genotyping or sequencing quality. Therefore, tests of Hardy Weinberg Equilibrium should only be performed and presented for non-deletion patients. If the authors only calculated this metric for non-deletion patients, it is not clear from the text.

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).

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.

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:
I declare that I have no competing interests.