Reviewer's report

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

Version: 1 Date: 4 June 2011

Reviewer: Sintia Belangero

Reviewer's report:

General comments:

Human gene names should be written in italic and all capital fonts (gene international nomenclature).

Title:

The word “discovery” should be changed for “detection”.

Abstract:

1) To define CTD acronym.
2) The authors have proposed to determine the pathogenesis of 22q11.2 del syndrome, however, no 22q11.2 del syndrome patient was included in this study. Only isolated conotruncal heart defects patients were recruited. To change pathogenesis of 22q11.2 del syndrome for pathogenesis of conotruncal heart defects.

Methods:

Unnecessary describing karyotype and MLPA protocols, since they are widespread and established techniques. This part can be drastically shortened.

Subheading “Gene PCR and Sequence” should be changed for “Gene PCR and Sequencing”.

Results:

In the first line: The authors should add “numaerical”, e.g. “No obvious structural and numerical abnormalities”

Regarding to subheading MPLA:

1) The whole first paragraph should not be in “Results”. This is a method specification.

3) When describing the deletion size (for example, CLTCL1-LZTR1), to quote in Mb and to give respect to LCRs which are well-known references.

Regarding to subheading FISH:

1) The whole first paragraph should not be in “Results”. This is a method specification.

2) To indicate where the D22S75 marker is located on region map (figure 2).
TBX1 Sequencing:
To change “a frequence of p>0.05” for “a p value>0.05”
The statements “It may, therefore, be implied that sister chromatid wich…” and “Prior studies have found that although the TBX1…” should be in Discussion and not in Results.
Tables:
Considering the tables III, IV and V, there are nine SNPs reported, whereas in the text eight are mentioned. The rs41298838 is not presented in table III. Is it a mistake typing?
Figure 3:
It is useless and adds nothing to the comprehension.

Level of interest: An article of importance in its field

Quality of written English: Needs some language corrections before being published

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'below.