Reviewer's report

Title: Novel TBX1 loss-of-function mutation causes isolated conotruncal heart defects in Chinese patients without 22q11.2 deletion

Version: 3
Date: 5 May 2014

Reviewer: Maria Cristina Digilio

Reviewer's report:

Minor essential revisions

The authors report on a novel de novo TBX1 loss-of-function mutation as a cause of isolated conotruncal heart defect in a Chinese patient without 22q11.2 deletion.

The report is noteworthy as short observation, but more clinical details regarding the mutated subject should be added.

Clinical criteria of inclusion of patients are weak (the authors state that patients had CHD without deletion 22q11.2 and no extracardiac anomalies were present. Which kind of extracardiac anomalies were absent?

Can the authors provide clinical details of facial appearance of the mutated patient? I realize that it is probably difficult to obtain, but a photo of the face of this patient should be useful.

How was excluded mild cognitive deficit?

More clinical details should be added before stating that the TBX1 mutation is a cause of “isolated” conotruncal defect.

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'