Reviewer’s report

Title: A novel TAB2 nonsense mutation (p.S149X) causing autosomal dominant congenital heart defects: a case report of a Chinese family

Version: 0 Date: 10 Nov 2019

Reviewer: George Makavos

Reviewer's report:

Interesting work on a novel TAB2 nonsense mutation linked to congenital heart defects. However there are some issues to be addressed by the authors in order to raise the competence of this article.

- The proband was tachypneic with occasional fatigue. Where these symptoms clearly associated to mild aortic stenosis and mild mitral, tricuspid and aortic regurgitation? It is not clear whether structural abnormalities of the aortic valve (such as bicuspid aortic valve) were detected by transthoracic echocardiogram.
- High prevalence of mild mitral and tricuspid regurgitation have been reported in the general population worldwide, however not associated with clinically significant symptoms and signs of heart failure.
- It is not addressed whether any screening for other causes of tachypnea was performed (anemia, respiratory infection, pulmonary embolism e.t.c) or tachypnea was solely associated to pregnancy and valvular heart defects.
- Left coronary artery dilation in proband's relatives were detected by transthoracic echocardiogram or other imaging modalities? (coronary angiography, coronary CT, angiography). Since transthoracic echocardiogram has low sensitivity for the identification of coronary aneurysms, it would be informative to address by which imaging technique coronary abnormalities were detected in relatives.
- It would be constructive to add comments in the discussion segment regarding any potential association of the TAB2 mutation with sudden cardiac death during childhood or adulthood.

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

Yes

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

Yes

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?

Yes
If an additional statistical review is recommended, please specify what aspects require further assessment in your comments to the editors.

I am able to assess the statistics

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