Reviewer’s report

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

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Reviewer: Alexandra Frogoudaki

Reviewer's report:

A novel TAB2 nonsense mutation (p.S149X) causing autosomal dominant congenital heart defects: a case report of a Chinese family is a very interesting paper describing a novel TAB2 mutation that causes heart defects.

There are some comments

1. Atrial septal aneurysm is not a valvular heart disease
2. Authors describe very mild valvular disease. Mild mitral, tricuspid and pulmonary regurgitation could be a relatively normal finding. Authors should check patients records and state if regurgitation of the above mentioned valves in cohort patients are more than expected;
3. The patient III:7 died at the age of 2 years old by sudden cardiac death (SCD). She had left and right ventricular dilatation. Is there any information concerning arrhythmias or ejection fraction? Could TAB2 mutation be attributed to SCD and in what way? Authors should discuss this important issue.

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.

Unable to assess

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

Not relevant to this manuscript

Quality of written English
Please indicate the quality of language in the manuscript:
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