Reviewer’s report

Title: Atrial septal defect (ASD) in a patient with congenital disorder of glycosylation type 1a (CDG-1a): a case report

Version: 0 Date: 29 Jun 2017

Reviewer: Divyanshu Mohananey

Reviewer's report:

1. Do you believe the case report is authentic?

Yes/No

While I believe the authors have followed rigorous genetic testing protocols to confirm CDG 1 mutation along with enzymatic changes of PMM2 activity, I am not convinced the ASD can be ascribed to CDG1. ASD is a fairly common congenital cardiac disorder with close to 5-6 cases per 100,000 live births. In such a scenario can it not be a possible that both these conditions co-existed in the presented patient? I would be weary of rushing to causality and association in this situation. I feel the authors should tone down their conclusions about this being the first ever reported case of CDG1 leading to ASD.

2. Do you have any ethical concerns? Please consider if local Institutional Review Board approval or ethical approval was obtained (if appropriate) and if the patient (or their parent or guardian in the case of children under 18) gave written, informed consent to publish this case and any accompanying images. A statement to this effect should appear in the manuscript.

Comments:

No mention of consent from parents. The authors only mention that they took consent for genetic testing. Consent for presenting this report to this journal is also warranted.

3. Does the Introduction explain the relevance of the case to the medical literature?

Yes/No

Yes
4. Does the article report the following information? Where information is missing, please specify.

a. The relevant patient information, including:
   - De-identified demographic information (age, gender, ethnicity)
   - Main symptoms of the patient
   - Medical, family and psychosocial history
   - Relevant past interventions and their outcomes
   Well presented case with follow up information

b. The relevant physical examination findings

c. Important dates and times in this case (if appropriate, organized as a timeline via a figure or table); if specific dates could lead to patient identification, consider including time relevant to initial presentation, i.e. initial presentation at T = 0, follow up at T = 1 month.

I believe we would benefit if a table were presented with symptoms, physical findings and diagnostic testing.

d. Diagnostic assessments, including:
   - Diagnostic methods
   - Challenges (e.g., financial, language/cultural)
   - Reasoning and prognostic characteristics (e.g., staging), where applicable

See above

e. Types and mechanism of intervention

More detailed description of treatment, especially for the ASD should be provided. Also, follow up echocardiograms if any should be mentioned with timeline.

f. A summary of the clinical course of all follow-up visits
Comments:

As above

5. Is the interpretation (discussion and conclusion) well balanced and supported by the case presented?

Comments:

See first point

6. Is the anonymity of the patient protected? Please consider any identifying information in images such as facial features or nametags, whether the patient is named etc. If not, please detail below.

Yes/No

Yes

7. Is the Abstract representative of the case presented?

Comments:

Yes

8. Does the case represent a useful contribution to the medical literature?

Comments:

It is no doubt an interesting case. However, as mentioned above I think the conclusion that ASD is a result of genetic mutations may be far fetched in this case. Moreover, the authors themselves concede that the mutations known to be associated with ASD were in fact absent in this patient.

9. Additional comments for the author(s)?

None
**Level of interest**
Please indicate how interesting you found the manuscript:

An article of limited interest

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

Not suitable for publication unless extensively edited

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Please complete a declaration of competing interests, considering the following questions:

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3. Do you hold or are you currently applying for any patents relating to the content of the manuscript?

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5. Do you have any other financial competing interests?

6. Do you have any non-financial competing interests in relation to this paper?

If you can answer no to all of the above, write 'I declare that I have no competing interests' below. If your reply is yes to any, please give details below.

None

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