Author's response to reviews

Title: Duplication of the ALDH1A2 gene in association with pentalogy of Cantrell: a case report

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Author's response to reviews: see over
Dear Editorial Team,

Thank you for your review of this manuscript entitled, “Duplication of the ALDH1A2 gene in association with pentalogy of Cantrell: a case report.” We have read with interest the comments provided by the reviewers and the editorial team, and the manuscript has been revised accordingly. Following, you will find a point by point description of the changes made. Changes have been made using MS Word’s track changes feature.

In response to the Editorial Team:

1. The title has been revised to include the study design. “: a case report” has been added to the title.
2. Co-authors’ emails have been added to the title page.
3. The patient’s age, sex, and ethnic background have been added to the Abstract: case presentation section. The brief details of what the patient presented with are also included.
4. The ethnicity of the patient (and mother) have been added to the Abstract and Case presentation section.
5. A list of abbreviations used in the manuscript has been added after the conclusions section. The abbreviation CDH was taken out of the conclusion section since it had not been used previously within the manuscript.

In response to Referee 1 (Colin J McMahon):

1. The last sentence in the case presentation section has been addended to read …inherited from a phenotypically normal mother with no known …defects. The final sentence in the conclusion has the word maternal added before retinoic acid levels to be clear on the idea that since the mother is an apparently normal carrier of the duplication found in the infant with pathology, we postulate that there is a possibility that alteration in maternal retinoic acid levels during gestation could have resulted in the observed fetal pathology.

In response to Referee 2 (Junji Takaya):

1. The discussion has been trimmed down and simplified by shortening the last sentence of paragraph 2, and eliminating paragraph 3. Secondary to this change, the original references [7] and [8] are removed and the remaining references are renumbered accordingly.
2. Arrows have been added to Figure 1. The arrows have been labeled a and b, and the a and b have been added to the figure legend.
3. Figure 2 has been removed from the manuscript since it would not be easily understood by most readers of this manuscript.

I appreciate the thoughtful responses provided by the reviewer. I think that the suggested changes strengthen this report, which represents, to our knowledge, the first genetic abnormality reported in association with Pentalogy of Cantrell.

Sincerely,

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