Reviewer's report

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

Version: 1 Date: 12 September 2013

Reviewer: Colin J McMahon

Which of the following following best describes what type of case report this is?: New associations or variations in disease processes

Has the case been reported coherently?: Yes

Is the case report authentic?: Yes

Is the case report ethical?: Yes

Is there any missing information that you think must be added before publication?: Yes

Is this case worth reporting?: Yes

Is the case report persuasive?: Yes

Does the case report have explanatory value?: Yes

Does the case report have diagnostic value?: Yes

Will the case report make a difference to clinical practice?: Yes

Is the anonymity of the patient protected?: Yes

Comments to authors:

Steiner et al. report a child with pentalogy of Cantrell and microduplication of chromosome 15q21.3. This is a novel finding and adds to our knowledge of this condition. Potential candidate genes within this region are discussed including the potential role of the ALDH1A2 gene. They postulate that perturbations in retinoic acid generation may be the mechanism underlying development of the pentalogy of Cantrell in this child.

Given that the mother had the same gene mutation, did you screen her for a diaphragmatic or sternal defect or presence of a septal defect?

The report is nicely written, and the conclusions and figures are clear to the reader.
Quality of written English: Acceptable

Declaration of competing interests:

I have no competing interests in relation to this paper.