Reviewer's report

Title: PITX2 mutations are not a common cause for transposition of the great arteries (dTGA)

Version: 1 Date: 22 October 2004

Reviewer: Martin Blum

Reviewer's report:

General

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Major Compulsory Revisions (that the author must respond to before a decision on publication can be reached)

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Minor Essential Revisions (such as missing labels on figures, or the wrong use of a term, which the author can be trusted to correct)

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Discretionary Revisions (which the author can choose to ignore)

Discretionary Revisions

I recommend two changes to improve the manuscript:
1. The amplified regions should be indicated in Fig. 1. Otherwise the mere reproduction of the gene locus and isoforms should be deleted, as it is available in the published literature.
2. The formal possibility that the transcript levels of Pitx2c were drastically reduced in TGA patients due to mutations in the promotor region of the gene should not be dismissed, but rather clearly be stated. It may indeed be worthwhile to reexamine the Pitx2c promotor region of the 96 samples in order to unequivocally rule out an involvement of this gene in human TGA.

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What next?: Accept after discretionary revisions

Level of interest: An article of importance in its field

Quality of written English: Acceptable

Statistical review: No

Declaration of competing interests:
none