Reviewer’s report

Title: Cascade Fumarate Hydratase mutation screening allows early detection of kidney tumour: a case report.

Version: 0 Date: 06 Dec 2016

Reviewer: Aikseng Ooi

Reviewer's report:

HLRCC is a rare hereditary cancer syndrome that is typically diagnosed by dermatologists whereby patients present with cutaneous leiomyomas. In this manuscript, Melanie Chan and coworkers report an interesting case study of a HLRCC family whereby the diagnosis was made because their newborn daughter was diagnosed with fumarate hydratase deficiency. The diagnosis prompted renal ultrasonography screening, which lead to the early detection of a kidney lesion in the otherwise asymptomatic father. This is particularly significant because HLRCC associated kidney cancer is very aggressive and is refractory to all known treatment. The early detection saved the father's life and this case study will help cultivate awareness among health care providers.

Minor concerns:

1. If possible electropherograms or equivalent diagram from the mutation screen should be shown.

2. Typographical and terminological mistakes at Line 139, "… hypothesised that fumarate may act as an 'oncoprotein' in FH-deficient kidney cancer." Should be hypothesized that fumarate may act as an oncometabolite in FH-deficient kidney cancer.

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.

Yes

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:

Acceptable

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