Reviewer's report

Title: Paraganglioma and pheochromocytoma upon maternal transmission of SDHD mutations

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Reviewer: Takeshi Usui

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The manuscript showed the three cases of “apparently maternally transmitted” SDHD related paraganglioma. The case 3 had been concludes as a photocopy of PGL (sporadic paraganglioma in an SDHD mutation carrier).

In case 2, although immunohistochemical analysis may suggest the pathogenity of SDHx, the authors could not demonstrate the LOH in adrenal medullary tissue (as the paraganglioma tissues were not available). Therefore, authors could not conclude that the paragangliomas in this case arise from the maternally transmitted Asp92Tyr SDHD mutation itself. As this mutation is reported as a founder mutation in Netherlands, they must be very careful about the pathogenity of this mutation in Netherland. The authors should discuss this point.

In case 1, the authors describe that “maternal family of this patient had history of paraganglioma”. Who are the patients in this family? Please indicate the affected members in Fig 1a. Also, the authors should describe the precise clinical phenotype of the parents of case 1. The data of LOH is ambiguous by both Sanger sequence analysis and microsatellite analysis (Fig 2a, b). Both data indicate incomplete LOH in the tumor. Please discuss this point (contamination of normal tissue?). The tissue should be obtained by laser captured micodisection to avoid contamination of normal tissue. In Fig 2b and 2c, the authors should show all the profiles of microsatellite markers alleles.

Level of interest: An article whose findings are important to those with closely related research interests

Quality of written English: Acceptable

Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:

I declare that I have no competing interests.