Author’s response to reviews

Title: Pheochromocytoma as a rare cause of hypertension in a 46 X, i(X)(q10) Turner syndrome: a case report and literature review

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Reviewers’ comments

Reviewer 1:

Comment 2

I consider that it should be mentioned that genetic testing was proposed to the patient on grounds of early age of presentation, in accordance with relevant guidelines. It is thought immense to ask if patients with Turner syndrome are genetically more susceptible to pheochromocytoma, considering the scarceness of data.
Thank you for your kind review and comments. I added this point in discussion section as below (page 9, line199).

Pheochromocytomas are known to be hereditary in 30-40% of cases, and hereditary catecholamine-secreting tumors typically present at a younger age than sporadic tumors [24]. Genetic testing should be considered in all patients and is strongly indicated in specific patients such as those with unilateral adrenal pheochromocytoma onset at a young age, those with a positive family history of pheochromocytoma and paraganglioma or carriers of tumor susceptibility gene mutations, and those with syndromic features or metastatic disease [19]. However, unfortunately, our patient had no family history of pheochromocytoma and she refused a genetic testing due to expensive cost of genetic testing. Further studies including genetic tests are necessary to define the mechanisms underlying association between Turner syndrome and pheochromocytoma.

Comment 5

I would like to add that MIBG demonstrates false-positive results in pheochromocytomas, with up to 50% of normal adrenal glands showing physiological uptake, often asymmetrical. Perhaps MRI should be considered in lieu of MIBG.

Reply 5)

Thank you for your kind comments.

MIBG scintigraphy is superfluous in patients with sporadic solitary adrenal pheochromocytoma identified on CT or MRI. However, in our case, CT scan had a limited ability for diagnosis for typical pheochromocytoma due to relative small size of tumor. Therefore, we performed MIBG scan which had a high specificity for diagnosis of pheochromocytoma and for detection of extra-adrenal pheochromocytoma in view of young age.

Comment 7

Repeating urine measurement in a second sample could have been considered, due to unusual biochemical profile.

Reply 7)

Thank you for your kind comments.