



PP-A-10

RARE AND AGGRESSIVE METASTATIC PHEOCHROMOCYTOMA RECURRENCE IN A YOUNG ADULT MALE WITH MEN 2A SYNDROME

<https://doi.org/10.15605/jafes.037.AFES.28>

Edrome Hernandez

Philippine General Hospital, Manila, Philippines

BACKGROUND

Multiple Endocrine Neoplasia 2A (MEN 2A) is characterized by 70-100% risk of medullary thyroid cancer, 50% risk of pheochromocytoma, and 20-30% risk of primary hyperparathyroidism mainly caused by RET mutation. Pheochromocytoma in MEN 2A is almost always benign and often follows an indolent course.

CASE

We demonstrate the clinical course and challenges in the management of a case of metastatic pheochromocytoma recurrence in a patient with MEN 2A.

We present a 31-year-old male with histopathologically-confirmed bilateral pheochromocytoma occurring metachronously. He was subjected to left adrenalectomy and right adrenalectomy in 2014 and 2020, respectively. The patient was also diagnosed with primary hyperparathyroidism and medullary thyroid carcinoma presenting with mild hypercalcemia and multiple thyroid nodules. He underwent total thyroidectomy with neck dissection and left inferior parathyroidectomy. Genetic testing revealed the presence of a RET mutation. One year post-surgery, the patient had persistent elevation of 24-hour urine metanephrines [13.977 mg (NV: 0-1)]. Adrenal CT demonstrated a 2.1 cm ovoid focus in the right suprarenal region and I-131 MIBG scan showed avid uptake on the right frontal bone. Patient underwent excision of right adrenal bed and right frontal bone tumor which were confirmed to be metastatic pheochromocytoma on histopathologic report. He achieved normalization of 24-hour urine metanephrines 0.8 mg (0-1 mg) one month postoperatively.

CONCLUSION

Metastatic pheochromocytoma is only seen in less than 4% of MEN 2A. Metastatic PHEO recurrence has been reported only in few case reports and optimal management of these cases requires a multi-specialty approach.

PP-A-11

PHAECHROMOCYTOMA IN A PATIENT WITH SUSPECTED BIRT-HOGG-DUBÉ SYNDROME: AN INCIDENTAL OR COMPOSITE RELATIONSHIP?

<https://doi.org/10.15605/jafes.037.AFES.29>

Rabiu Momoh, Alamin Alkundi, Marcus Nash

William Harvey Hospital, Ashford, Kent, United Kingdom, Ashford, United Kingdom

BACKGROUND

A rare case of pheochromocytoma in a 57-year-old female patient with a provisional clinical diagnosis of Birt-Hogg-Dubé (BHD) syndrome is presented. Whether this is an incidental finding or whether a causative or composite relationship between these two entities remains to be fully described. As much as we are aware, our case would be the second of such association to be described in literature.

CASE

The patient's hospital record was reviewed after consent was obtained.

Bilateral renal cysts and a right adrenal lesion were first identified in the patient at age 48 from abdominal CT studies done for her recurrent bilateral flank pain. To date, two percutaneous drainages of cysts on the right kidney were done to relieve pain. Following assessment of patient at a tertiary hospital's genetics clinic at age 52, a provisional diagnosis of Birt-Hogg-Dubé (BHD) syndrome was made for her bilateral renal cysts and notable skin lesions. Her CT chest study did not reveal lung cysts. Genetic mutation studies for FLHN, FH, MET, SDHB and VHL genes were negative. The patient's right adrenal lesion was under surveillance and eventually required right adrenalectomy for newly developed recurrent episodic hypertension at age 54. Histology showed paraganglioma-like appearance, a 2% Ki-67 index and a negative SDHB staining. The patient's daughter was also diagnosed with bilateral renal cysts and recurrent spontaneous pneumothoraces and a provisional diagnosis of DHB syndrome variant was also made for her.

CONCLUSION

Further research work into the above rarely-described clinical association needs to be done.