PA-A-10

FIRST CASE OF EXCLUSIVELY DOPAMINE-SECRETING PARAGANGLIOMA IN MULTIPLE ENDOCRINE NEOPLASIA TYPE 2A (MEN2A)

https://doi.org/10.15605/jafes.037.S2.16

Chiew Ken Seng, Velaiutham Shanty, Khaw Chong Hui
Hospital Pulau Pinang, Penang, Malaysia

INTRODUCTION
Pheochromocytomas in MEN2A are usually intra-adrenal, though they may uncommonly manifest as paragangliomas. Predominantly or exclusively dopamine-secreting pheochromocytomas and paragangliomas (PPGL) are rare with only 33 cases reported in the literature. We report the first case of exclusively dopamine paraganglioma in the context of MEN2A.

CASE
A 72-year-old male was diagnosed with MEN2A following a family screening in 1996. Genetic analysis revealed a mutation in codon 634 of the RET proto-oncogene (C634Y). He underwent total thyroidectomy for medullary thyroid carcinoma in 1996 and total parathyroidectomy for primary hyperparathyroidism in 1997. His yearly 24-hour urinary catecholamines had been within the normal ranges. However, in August 2019, his urinary dopamine was raised to 1033 µg/day (Normal range: 64.0-400). Urinary adrenaline and noradrenaline were not elevated. Repeated 24-hour urinary metanephrines in August 2020 yielded an elevated 3-methoxytyramine level of 21.8 µmol/day (Normal range: 0.10-1.79). Urinary metanephrines and normetanephrines remained within normal ranges. He has hypertension which was well-controlled on two agents. He is, otherwise, asymptomatic with no paroxysmal attacks of headaches, sweating or palpitations. Iodine-131 meta-iodobenzylguanidine (I-131 MIBG) imaging revealed an avid lesion in the mediastinum with no tracer uptake at the adrenal glands. The patient declined further interventions.

CONCLUSION
Despite the rarity of exclusively dopamine-secreting PPGL, the case highlights the importance of measuring urinary or plasma dopamine in MEN2A. Dopamine-secreting PPGL typically lacks the classical presentation of paroxysmal attacks and is often extra-adrenal. The patient was diagnosed with PPGL at 70 years of age. As the prevalence of PPGL in MEN2A increases with age, there is no age cut-off to stop screening.

PA-A-11

RARE ASSOCIATION OF NEUROFIBROMATOSIS TYPE 1 WITH PRIMARY HYPERPARATHYROIDISM: A CASE REPORT

https://doi.org/10.15605/jafes.037.S2.17

Melody Tsen Shu Ling,[1] Mohd Idris Bin Mohd Diah,[1] Tee Hwee Ching[2]
[1]*Hospital Tawau, Sabah, Malaysia*
[2] *Hospital Queen Elizabeth II, Sabah, Malaysia*

INTRODUCTION
Neurofibromatosis Type 1 is an autosomal dominant genetic disorder characterized by central nervous system involvement, cutaneous manifestations and increased risk of developing endocrine-related tumors. Meanwhile, Primary Hyperparathyroidism is most commonly caused by parathyroid adenoma resulting in abnormal calcium homeostasis. There are case reports identifying an association between neurofibromatosis and primary hyperparathyroidism as a variant of multiple endocrine neoplasia (MEN) syndrome, however, their association is not fully understood.

CASE
This is the case of a 50-year-old male patient who is known to have neurofibromatosis type 1 with cutaneous manifestations (cafe au lait spots, neurofibromas, Lisch nodules) and pleural neurofibroma. He initially presented with persistent left lumbar pain and a kidney ultrasound demonstrated evidence of left nephrolithiasis. Laboratory tests revealed high serum calcium of 2.7 mmol/L to 3.1 mmol/L, a low phosphate level of 0.49 mmol/L and a markedly raised serum parathyroid hormone level of 1038 pg/ml. Vitamin D level was normal at 72.26 nmol/L and a calcium-to-creatinine-ratio of 0.019. Further workup with Sestamibi parathyroid scan revealed evidence of parathyroid adenoma inferior to the left thyroid gland. Contrast-enhanced computed tomography of the neck showed a heterogeneously-enhancing lesion in the same location corresponding with the Sestamibi scan finding. There was no clinical evidence of pheochromocytoma and the 24-hour urine metanephrines test was normal. With the imminent complication of nephrolithiasis, the patient is awaiting parathyroidectomy.

CONCLUSION
This case demonstrates a rare but proven co-occurrence between neurofibromatosis and primary hyperparathyroidism as evidenced by biochemical tests and radiographic imaging. Screening neurofibromatosis patients for primary hyperparathyroidism during the initial evaluation and follow-up is a potential step for early detection of the condition before it leads to complications.