EP_A120

PRECISION MANAGEMENT IN A CASE OF PHEOCHROMOCYTOMA CATALYSING EARLY DETECTION OF MEDULLARY THYROID CARCINOMA

https://doi.org/10.15605/jafes.039.S1.131

Vanusha Devaraja, Azraai Bahari Nasruddin, Vijaya Mala Valayutham, Zanariah Hussein
Endocrine Unit, Department of Internal Medicine, Hospital Putrajaya, Malaysia

INTRODUCTION/BACKGROUND

Approximately 35% of pheochromocytoma carry a germline mutation, highlighting the importance of genetic screening in early detection and follow-up of patients with hereditary syndromes. We report a case of a young female in whom individualized pheochromocytoma management expedited the diagnosis of medullary thyroid cancer.

CASE

A 21-year-old female student presented to the intensive care unit with malignant hypertension complaining of severe headache and abdominal pain. Computed tomography (CT) of the abdomen revealed a heterogenous mass measuring 5.9 x 6.3 x 6.2 cm arising from the left adrenal gland. Plasma free normetanephrines were significantly elevated at 53 nmol/L (normal range: <0.9 nmol/L). She was prepared preoperatively with phenoxybenzamine and propranolol, and successfully underwent open left adrenalectomy. Intraoperative findings revealed a well encapsulated adrenal tumour measuring 7 x 6 cm, weighing 122 grams, confirmed as pheochromocytoma. Genetic testing revealed RET proto-oncogene missense mutation (Cys634Trp). Screening for medullary thyroid carcinoma (MTC) and primary hyperparathyroidism were carried out in order to exclude other possible coexisting disorders in MEN2A syndrome. Serum calcium and parathyroid hormone were normal. Neck ultrasound revealed TIRAD 4 nodules on the upper pole of the right and left thyroid lobe with the largest measuring 1.1 cm and 1.3 cm, respectively. Lymph nodes were not enlarged. Serum calcitonin was elevated at 111 ng/L (normal range: ≤7.6 ng/L). Fine needle aspiration of bilateral thyroid nodules revealed papillary thyroid carcinoma (Bethesda V). She subsequently underwent total thyroidectomy. However, pathological examination of the tumours disclosed low grade multifocal MTC, staining positive for calcitonin, chromogranin A, synaptophysin and CD56. There was no evidence of extrathyroidal tumour extension or lymph node metastasis.

The patient was started on replacement L-thyroxine post-operatively.

CONCLUSION

Early diagnosis of multifocal MTC had been made in this young female due to the benefit of germline mutation. This has reduced the likelihood of unfavorable clinical outcomes associated with MEN2 syndrome.

EP_A121

HARMONISATION OF MACROPROLACTIN REPORTING

https://doi.org/10.15605/jafes.039.S1.132

Pavai Sthaneshwar,[1] Subashini C. Thambiah,[2] Jeyakantha Ratnasingam[3]
[1]*Department of Pathology, Faculty of Medicine, University Malaya, Malaysia*
[2]*Department of Pathology, Faculty of Medicine, University Putra Malaysia*
[3]*Endocrine Unit, Department of Medicine, Faculty of Medicine, University Malaya, Malaysia*

INTRODUCTION/BACKGROUND

The post-analytical phase is the last phase of the total testing process and it is an important step since it maximises the quality and effectiveness of laboratory information. Macroprolactin is a well-known analytical problem in laboratory diagnostics. There is no agreement on when to screen for macroprolactin in patients with hyper-prolactinaemia nor is there consensus on the reporting, terminology and reference intervals for macroprolactin.

METHODOLOGY

To create a consensus document that standardises the reporting of prolactin results after precipitation with polyethylene glycol (PEG) to minimise errors in result interpretation.

As a part of the joint initiative by the Chapter of Chemical Pathology and Metabolic Medicine, College of Pathologists, Academy of Medicine Malaysia and the Malaysian Association of Clinical Biochemists to harmonise the laboratory reporting of macroprolactin in Malaysia, an audit was conducted to obtain feedback from endocrino-logists from public, private and academic institutions on macroprolactin reporting. The questionnaire included the level of prolactin that was considered as hyperprolactinaemia, its cut-off value in males and female and whether screening for macroprolactin should be clinician-centred or laboratory-centred. It also addressed post-PEG precipitation, whether the report should specify the presence of macroprolactin and the preferred reporting format.