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

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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

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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.

RESULTS
Responses were obtained from 33 endocrinologists. The majority (81%) used reference limits given rather than a fixed cut-off of 700 m IU/L and 70% used a sex-specific range. More than half (69%) favoured that the laboratory should screen for macroprolactin in all samples with high prolactin. On report format, 61.5% preferred the use of post-PEG monomeric reference ranges and 100% required the laboratory to indicate the presence of macroprolactin.

CONCLUSION
The laboratory working group will consider the responses from endocrinologists in preparing the consensus recommendations on reporting macroprolactin and the reference intervals.

EP_A122

UNVEILING PITUITARY PARADOX: OCTREOTIDE LONG-ACTING RELEASE (LAR) INDUCED APOPLEXY IN POST-OPERATIVE RESIDUAL MACROADENOMA

https://doi.org/10.15605/jafes.039.S1.133

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INTRODUCTION/BACKGROUND
The development of a somatostatin analogue (SSA) has revolutionized the treatment of acromegaly. Octreotide LAR is a long-acting release formulation of SSA, often used as an alternative when surgery is not possible in patients with acromegaly or as an adjuvant therapy in post-operative patients with residual pituitary adenoma. Pituitary apoplexy, characterized by infarction or bleeding of the pituitary gland, is a rare condition.

CASE
An 18-year-old male with a clinical and biochemical diagnosis of acromegaly underwent transsphenoidal surgery in November 2021 with incomplete tumour resection, complicated by pituitary apoplexy after administration of octreotide LAR.

Post-operatively, he had residual pituitary adenoma with optic chiasm compression and persistent elevations of insulin-like growth factor 1 (IGF-1) (1.7x > upper limit of normal) and growth hormone (GH) level (10x > normal limit). After extensive discussion, the patient was started on medical treatment, cabergoline 0.5 mg twice weekly, but failed to achieve biochemical control despite continued use for 9 months. Subsequently, octreotide LAR 30 mg monthly was started, aiming to achieve better biochemical control and shrink the tumour size while awaiting stereotactic radiosurgery.

Unfortunately, 6 weeks after the first injection of octreotide LAR, he developed a sudden severe headache and visual disturbance, presenting clinically with bitemporal hemianopia, subsequently diagnosed with pituitary apoplexy on cranial MRI. Octreotide LAR was discontinued. Patient underwent repeated transsphenoidal surgery, which was uncomplicated, albeit with pre-existing central hypothyroidism and hypocortisolism.

CONCLUSION
Pituitary apoplexy is one of the rare complications of SSA. However, clinicians need to maintain a high level of suspicion for this complication if the patient presents with sudden headache with or without neuro-ophthalmic signs after receiving SSA, given its significant morbidity and potential fatality.

EP_A123

A RARE CASE OF ECTOPIC GIANT PROLACTINOMA MIMICKING CLIVAL CHORDOMA

https://doi.org/10.15605/jafes.039.S1.134

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INTRODUCTION/BACKGROUND
Ectopic pituitary adenomas are extremely rare tumours which develop outside the sella turcica. The most common locations are the sphenoid sinus, clivus, suprasellar space, nasopharynx and cavernous sinus. Due to their rarity, these tumours are frequently misdiagnosed as other skull lesions such as chordoma, chondrosarcoma, meningioma or astrocytoma. We present a case of ectopic giant prolactinoma mimicking as clival chordoma.

CASE
A 38-year-old nulliparous female presented with oligomenorrhoea since the age of 20 and secondary amenorrhea at the age of 38 associated with galactorrhoea. Otherwise, she denied headache, symptoms of increased ICP or blurring of vision. There were no other symptoms to suggest pituitary hyper- or hypofunction. Her initial hormonal work-up showed hyperprolactinemia (>8000 ng/mL) with hypogonadotropic hypogonadism. MRI of the brain was signed out as clival chordoma measuring 2.3 x 3.6 x 4.1 cm displacing the pituitary gland and