中文題目：異位性庫欣氏症候群同時合併疑似甲狀腺髓質癌為其可能之促皮質素分泌來源

英文題目：Ectopic Cushing’s Syndrome Concomitant with Suspected Medullary Thyroid Carcinoma as the Possible Origin of Corticotropin Secretion

作　　者：陳冠亘，林奕廷，李美月，林昆德，洪薇雯

服務單位：高雄醫學大學附設醫院內科部內分泌新陳代謝科

Introduction

Ectopic Cushing’s syndrome(ECS) accounts for about 20% of adrenocorticotropic hormone (ACTH)-dependent Cushing's syndrome. As a presentation of paraneoplastic syndromes, ECS was mostly reported to be caused by small cell lung carcinoma, bronchial carcinoid, or thymoma. However, extrathoracic sources of neuroendocrine tumors are reported causing a challenging task in the diagnosis and management of ECS. Here we reported a case of ECS and discussed the diagnostic difficulties.

Case presentation

The 63-year-old female presented with progressive facial swelling, generalized edema, symmetric proximal limbs weakness and body weight gain for one year. She was diagnosed hypertension and diabetes mellitus about one year ago but both were not well-controlled with medications. The patient’s body mass index was 25.5 kg/m^2. Physical examination identified central obesity, moon face, buffalo hump, purple striae, pitting edema on four limbs, symmetric proximal limbs weakness and grade 2 goiter. Laboratory data revealed hypokalemia (3.0 mmol/L), hyperglycemia (242 mg/dl) and high hemoglobin A1C (8.6%). Plasma ACTH and cortisol concentrations were 90.2 pg/mL and 27 μg/dL in the morning, and 95 pg/mL and 31 μg/dL in the afternoon respectively, indicating the loss of normal diurnal rhythm of ACTH and cortisol secretion. Due to high ACTH level, sella MRI performed but without pituitary tumor identified. The plasma cortisol concentrations were 27.9 μg/dL after high dose dexamethasone suppression test, supporting the diagnosis of ECS. Further evaluations for ectopic ACTH secretion were done including chest and abdomen computed tomography, and bronchoscopy. As thyroid nodules found, fine-needle aspiration was arranged and the cytology showed atypia. Then elevated plasma calcitonin concentration (30.4 pg/ml) was noted. ECS due to medullary thyroid carcinoma was highly suspected which would be confirmed with pathology after total thyroidectomy.

Discussion

ECS is an uncommon entity among the rare disease of Cushing's syndrome. The origin of ECS can pose a diagnostic challenge when encountering an occult tumor. Early diagnosis and localization of the possible malignancy would be crucial to facilitate the surgical excision of the tumor, in the hope to prevent morbidity and mortality due to hypercortisolism-related complications.