CONTINUING MEDICAL EDUCATION ΣΥΝΕΧΙΖΟΜΕΝΗ ΙΑΤΡΙΚΗ ΕΚΠΑΙΔΕΥΣΗ

Internal Medicine Quiz – Case 21

A 67-year-old male was admitted to the hospital with complaints of diarrhea (10-15 stools per day), fatigue and unintentional weight loss of 10 kg over the past 12 months. During this period, the patient had been thoroughly investigated for intestinal pathology. An extensive stool work up for infectious causes (bacteria, parasites and Clostridium difficile) was negative. Repeated endoscopies performed mainly because of a very high serum value of carcinoembryonic antigen (CEA) (550 ng/mL, upper limit 5 ng/mL for smokers) were also negative. On admission the patient was anemic, with severe wasting syndrome. Apart from a palpable nodular lesion on the left thyroid lobe, the rest of the clinical examination was unremarkable. Laboratory examinations revealed severe anemia (Ht: 20.9%, Hb: 7.0 g/ dL, RBC: 1, 91×10⁶/L), while thyroid function tests were within normal limits. Whole body computed tomography (CT) detected a thyroid nodular lesion (fig. 1), as well as multiple hypodense liver lesions. Serum calcitonin determination revealed a very high value (11.816 ng/L, upper limit 11.7 ng/L).

Comment

Serum calcitonin is part of the workup of chronic diarrhea with no obvious cause, since persistent diarrhea may be the initial clinical feature in 30% of medullary thyroid carcinoma cases.\(^1\) Medullary thyroid carcinoma (MTC) occurs either as sporadic (75%) or in a hereditary form (multiple endocrine neoplasia type 2, MEN 2) due to germ line mutations in the RET proto-oncogene. Like most thyroid carcinomas, it may be asymptomatic for long; however,

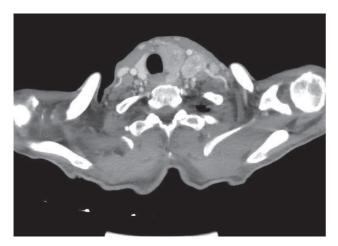


Figure 1. Computed tomography image (axial view) shows enlargement of the left thyroid lobe, where a solitary nodule with a heterogeneous contrast enhancement is detected.

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K. Kaziani,1

A. Assimakopoulou,²

P. Iliakis,1

G. Kassi²

¹Third Department of Internal Medicine, "Sotiria" Regional Hospital for Thoracic Diseases, National and Kapodistrian University of Athens, Athens ²Endocrinology Unit, "Sotiria" Regional Hospital for Thoracic Diseases, National and Kapodistrian University of Athens, Athens, Greece

large thyroid cancers can cause neck discomfort, dysphagia or hoarseness. Early local metastases to adjacent muscles, lymph nodes or the trachea are usually present at diagnosis. Eventually, late metastases may appear in the bones, lung, adrenals or liver. Ultrasonography of the neck is indicated as screening imaginary tool. For patients with local lymph node metastases on ultrasound or with high preoperative serum basal calcitonin further imaging is required to assess for metastatic disease. Thyroid function tests are usually normal. When MTC is suspected, biochemical evaluation for coexisting tumors (especially pheochromocytoma) must be performed, as well as serum CEA measurement, the levels of which are related to the tumor mass or disease dissemination. Genetic analysis is mandatory for first degree relatives. MTC is best treated with surgery for the primary tumor and metastases with a 10-year survival of 78%.^{2.3}

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Corresponding author:

K. Kaziani, Third Department of Internal Medicine, "Sotiria" Regional Hospital for Thoracic Diseases, National and Kapodistrian University of Athens, Athens, Greece e-mail: katkaziani@hotmail.com

Diagnosis: Medullary thyroid carcinoma