Diagnostic dilemma for an unusual presentation of thyroid malignancy

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ABSTRACT

Diagnosis is a crucial element for prompt identification and treatment of serious diseases. Thyroid malignancy presenting as brain metastasis is a rare entity while presenting as colon metastasis is extremely rare. Here, we discuss the difficulty faced with the diagnosis of the same. We report the case of a 66-year-old male, who was a known diabetic and hypertensive and presented with the complaint of the right facial nerve weakness. On evaluation, he was found to have an extradural lesion in the right petrous bone. He underwent pre/retrosigmoid craniotomy and subtotal decompression of lesion. Histopathology showed metastatic adenocarcinoma. He underwent positron emission tomography/computed tomography scan, which showed metabolically active foci in the posterior cranial fossa, ascending colon, manubrium sterni, and lung. Colonoscopy demonstrated a polyp in the ascending colon, biopsy of which revealed metastasis from the thyroid. Later, he was found to have a nodule of 1 cm in the right lobe of thyroid. Total thyroidectomy was done and the histopathology showed a multifocal papillary carcinoma thyroid follicular variant with tall cell areas and capsular invasion.

Key words: Carcinoma thyroid, Colon polyp, Facial palsy, Immunohistochemistry, Positron emission tomography scan

he frequency of differentiated thyroid carcinoma (DTC) initially presenting as distant metastatic disease accounts for 1%–9%. The most common site of metastasis for DTC is lung and bone [1]. Diagnosis is a crucial element for prompt identification and treatment of any disease. In malignancy, it becomes extremely difficult to treat when the clinical diagnosis of the primary is not known. This will necessitate a proper diagnostic investigation including radiological, histopathological, and immunohistochemical (IHC).

This case report highlights the importance of investigations necessary for proper and accurate diagnosis of the lesion, facilitating its management. Here, we discuss the difficulty faced with the diagnosis of a thyroid malignancy which prolonged definitive management. Few other salient features observed in this case includes thyroid malignancy presenting as facial nerve palsy with only a few cases reported, metastasis of malignancy to brain is rare and colon is extremely rare, and positron emission tomography (PET) negative thyroid malignancy, with uptake in distant metastatic sites.

CASE REPORT

A 66 years old gentleman had a history of facial weakness and reduced hearing from the right side for 8 months (Fig. 1). He was on regular medication for hypertension and diabetes. He was evaluated with magnetic resonance imaging scan, which showed a right petrous extradural mass lesion (Fig. 2). Subsequently, he underwent pre/retrosigmoid craniotomy and subtotal decompression for the same.

Histopathological examination from the lesion was suggestive of metastatic adenocarcinoma with diffusely positive thyroid transcription factor-1 (TTF-1) and vimentin negative. Positron emission tomography with fluorine-18 fluorodeoxyglucose integrated with computed tomography (18F-FDG PET/CT) showed metabolically active disease in the right posterior cranial fossa with the destruction of adjoining bone (SUV 9.3), concentration of FDG in soft tissue of midascending colon (SUV 27.97), expansile lytic-sclerotic lesion in manubrium sterni (SUV 8.95), and subcentimeter parenchymal nodule in lung (Fig. 3). Later on, the patient underwent a colonoscopy which showed a large ascending colon polyp (Fig. 4). Biopsy of the polyp was suggestive of metastatic adenocarcinoma.

The patient was referred to our institution for further management. The general clinical examination was normal including vitals. He had a clinically palpable 1×1 cm nodule in the right lobe of thyroid. IHC of the colonoscopy biopsy sample was consistent with metastasis from thyroid (thyroglobulin [TG] diffuse strong positive and TTF 1 strong positivity). TG level was 5457 ng/ml. Ultrasonography (USG) neck showed nodules in both lobes of the thyroid (Figs. 5 and 6).

Total thyroidectomy was done and the histopathology was reported as multifocal papillary carcinoma (thyroid follicular variant) with focal tall cell areas, largest nodule measuring $1\times0.8\times0.5$ cm with capsular invasion (Fig. 7). Following surgery, the patient underwent radioiodine ablation (30 millicuries) and was follow-up for the past 6 months.



Figure 1: Patient with the right facial nerve palsy

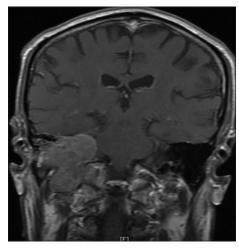


Figure 2: Magnetic resonance imaging showing lesion extradural lesion

DISCUSSION

The frequency of DTC initially presenting as distant metastatic disease amounts to 1%–9% [1]. Such cases represent a diagnostic challenge and require meticulous clinical, radiological, and histopathological evaluation to trace the primary. The most common site of metastasis for DTC is lung and bone. Our case being DTC with distant metastasis was a diagnostic challenge as it had metastatic lesion involving the uncommon sites such as brain and colon.

DTC has a good prognosis compared to other malignancies of the head and neck region, even when the cervical lymph nodes are involved. It has a drastic influence on survival and quality of life when there is a distant metastasis. Metastasis in the brain is noted in only about 1% of the all thyroid cases [2], and it occurs usually when multiple organs are involved. Involvement of the brain by DTC reduces the survival rate by 10 years depicting the grave nature of this disease. Metastasis of papillary carcinoma of the thyroid to colon or lower gastrointestinal tract is very rare and was first reported by Shaikh, in 2006 [2-4].

Clinical examination of such lesion should be done according to the case involved. If the thyroid gland is suspected, the

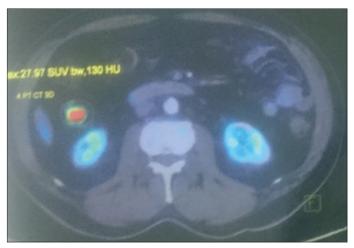


Figure 3: Positron emission tomography scan showing uptake in ascending colon



Figure 4: Colonoscopy showing polyp in colon

radiological investigation like USG of the neck or CT is used for the evaluation. FDG-PET/CT has no definitive consensus in the evaluation of DTC in routine clinical practice, as it might miss some details as most thyroid cancers are well-differentiated. In our case, there was PET uptake in distant metastasis site and no uptake in the thyroid. This might be either due to the small size of the thyroid lesion or dedifferentiation of the tumor in the distant site.

IHC plays an important role in the precise determination of the primary site of tumor origin. TG, TTF-1, and cytokeratin immunophenotype plays an important role in the diagnosis of probable site of tumor origin, in metastatic disease. TTF-1 is positive in malignancy from lung and thyroid, due to the similar embryological origin of tissues. TG immunoreactivity differentiates thyroid malignancy from lung malignancy. Here, IHC helped us to accurately determine the site of tumor origin, which further modified the management. Although thyroid malignancy was missed initially, IHC pointed out the right direction [5]. TG level is a specific, sensitive indicator of aggressive disease and predictor of relapse in DTC. Various studies have shown a sensitivity and specificity of 97% and 98%, respectively [6].

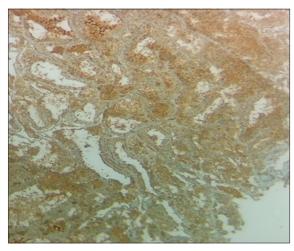


Figure 5: Thyroglobulin, immunostaining (colonic polyp)

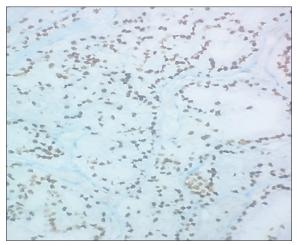


Figure 6: Thyroid transcription factor-1, immunostaining (colonic polyp)

To summarize, our patient presented with unusual metastatic disease initially, and the primary tumor was detected with the help of IHC of the metastatic specimen after a laborious workup. Thus, if IHC had been done completely, on brain lesion, we could have avoided all further investigations. IHC played a key role to arrive at the diagnosis, so proper selection IHC marker is important. Metastasis from thyroid should be considered as one of the differential diagnoses of the colonic polyp.

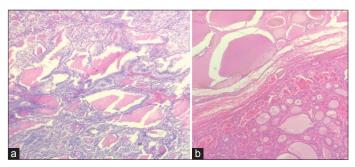


Figure 7: (a) Histopathology of polyp in colon (b) Histopathology of thyroid

CONCLUSION

Our case report points toward the unusual presentation of a thyroid malignancy that can lead to a lot of clinical/investigations related dilemma. Clinical examination can never be substituted, but IHC should be judicially used to complement clinical diagnosis. Brain and even colon lesion, which are suspected to be metastasis, should be evaluated thoroughly considering the possibility of metastasis from DTC, for better management of the patients.

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