Lhermitte-Duclos Disease Related With Cowden Syndrome Mimicking Metastatic Lung Cancer on FDG PET/CT

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Abstract: Cowden syndrome is characterized by multiple hamartomatous and neoplastic lesions including Lhermitte-Duclos disease, which is the main criterion for the diagnosis. Herein, we presented a patient with suspected metastatic disease referred to PET/CT, which showed mildly hypermetabolic multinodular thyroid goiter, multiple hamartomatous pulmonary, and breast nodules. Also, intense hypermetabolism was noted on the cerebellar tumor lesion. Lhermitte-Duclos disease was diagnosed based on the characteristic MRI findings, and she was followed up with a diagnosis of Cowden syndrome. Our case indicates that Cowden syndrome should be included as a differential diagnosis of abnormal FDG uptake in the multiple systemic hamartomatous tumors.

Key Words: Lhermitte-Duclos disease, Cowden syndrome, ^{18}F-FDG PET/CT

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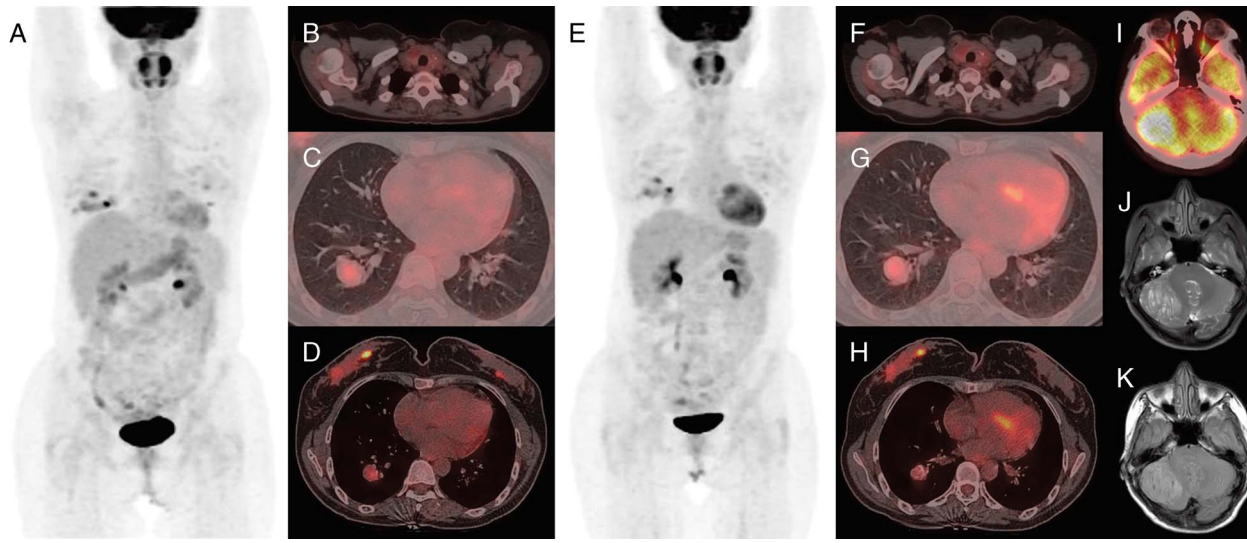

FIGURE 1. A 39-year-old woman with a medical history of colon polypectomy was referred to ^{18}F-FDG PET/CT scan for evaluating cancer of unknown primary due to multiple pulmonary nodules discovered on the thorax CT, which were evaluated in favor of metastases. MIP (**A**) and axial PET/CT images revealed mild-to-moderate hypermetabolic multinodular thyroid goiter (**B**) and multiple nodular lesions on both breasts (**D**). Increased FDG uptake was observed in multiple pulmonary nodules with calcification in both lung parenchyma, suggesting lung cancer (**C**). Also, intense FDG uptake (SUV_{max}, 13; tumor-to-contralateral cerebellum uptake ratio, 1.4) was depicted in the 5-cm measured hypodense mass lesion in the right cerebellar hemisphere suspicious for metastasis (**I**). The mass lesion showed tiger-striped pattern of hyperintensity on T2-weighted MR images without contrast enhancement (**J** and **K**), suggesting the diagnosis of Lhermitte-Duclos disease (LDD). The hamartomatous nature of the lesions along with LDD in the cerebellum lead to the diagnosis of Cowden syndrome (CS) and a year after, the tumors remained stable without a specific treatment (**E**, MIP; **F**–**H**, axial fused PET/CT images). LDD is a rare disorder characterized by a slowly growing hamartomatous tumor within the cerebellar cortex. Characteristic MRI findings including tiger-striped pattern on T2-weighted images and increased perfusion without contrast enhancement are pathognomonic for the diagnosis. Mild-to-moderate FDG uptake has been previously reported in LDD.[1,2] High FDG uptake in LDD may lead to misdiagnosis and unnecessary surgical procedures.[3] The mechanism of high FDG uptake in this benign tumor remains unknown but is sought to be related to the increased density of dysplastic cells.[4] Recent studies have revealed a close relationship between LDD and CS, also called multiple hamartoma-neoplasia syndrome, and adult-onset LDD is reported to be identical to CS.[5] CS is characterized by multiple benign tumors including mucocutaneous lesions, systemic hamartomas, gastrointestinal polyps, breast fibroadenomas, multinodular goiters, and increased risk of other neoplasms such as breast, thyroid, and endometrial cancers.[6] Whole-body FDG PET/CT has an emerging role in determining the presence/absence of cancer in CS.[7,8] Our case indicates that intense FDG uptake in such benign tumors including LDD can be misinterpreted as malignant disease; CS should be considered for correct interpretation of the PET activity to save patients from unnecessary surgery or chemotherapy.