Non-Small Cell Bronchogenic Carcinoma and Peutz-Jeghers Syndrome

To the editor:

In Peutz-Jeghers syndrome, multiple hamartomatous gastrointestinal polyps are associated with melanin pigmentation of the buccal mucosa, lips, hands, feet, and occasionally, the perianal region. Affected individuals have an increased risk of malignant tumors,[1] particularly gastrointestinal but also genital, mammary, and pancreatic tumors. However, few cases have been published in which Peutz-Jeghers syndrome is associated with non-small cell bronchogenic carcinoma. We describe such a case.

The patient, a 43-year-old man, was a smoker (18 pack-years) who had been diagnosed 20 years previously with Peutz-Jeghers syndrome and had been surgically treated 12 years previously for a benign mammary nodule in the right breast. The patient presented with cough, hemoptysis, and right-sided pleuritic pain that had persisted for 1 week. There were no other signs or symptoms, either thoracic or associated with other organs or systems. Physical examination revealed the presence of melanosis around the mouth, while other areas were found to be normal. Results of a hemogram, biochemical and coagulation analysis, and respiratory function tests were all normal. A chest radiograph revealed a mass in the right parahilar region and a more peripheral nodule, along with reduced volume of the upper right lobe. Computed tomography (CT) revealed a mass in the upper portion of the right hilum abutting the right pulmonary artery, the right main bronchus, and the upper lobar bronchus and its branches; a more peripheral nodule was also observed in contact with the pleura and diseased lymph nodes were observed in the right paratracheal area of the mediastinum, the largest being 2.5 cm in diameter. Fiberoptic bronchoscopy revealed an obstruction at the entrance of the upper lobar bronchus caused by tumor infiltration of the anterior wall and in the vicinity of the main bronchus; biopsy of the obstruction revealed large-cell bronchogenic carcinoma. CT of the abdomen did not reveal abnormalities. Bone scintigraphy was normal. Head CT with contrast enhancement revealed the presence of a single metastatic lesion in the right parietal region (stage IVc). Treatment with cerebral radiotherapy and chemotherapy was prescribed.[2]

Lung adenocarcinomas are associated with a high frequency of mutations (probably more than 30%) in the 2 copies of the *LKB1* gene.[3] These are somatic mutations; in other words, they are only present in the tumor and not in healthy tissue, and therefore, are not inherited. Patients with Peutz-Jeghers syndrome carry germline mutations in *LKB1*,[4] meaning that the mutations are already present in normal tissue, but only on 1 of the 2 chromosomes. When these patients develop tumors, including non-small cell bronchogenic carcinoma, they have 2 mutated copies of the gene.

Fiberoptic bronchoscopy did not reveal the presence of hamartomatous lesions, indicating that this primary tumor developed independently[5] and was associated with the genetic factors mentioned.

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