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Bronchioloalveolar Carcinoma in Peutz-Jeghers Syndrome: A Case Report

Peutz-Jeghers Sendromunda Bronkoalveoler Karsinom: Olgu Sunumu

Burcu Yormaz, Baykal Tülek, Mecit Süerdem

Abstract

Peutz-Jeghers syndrome (PJS) is a rare, autosomal dominant genetic disease. There is an increased prevalence of cancer in the PJS. A 48-year-old male patient with PJS was admitted to the clinic with bilateral consolidated areas in the thorax observed on a computed tomography image. A bronchoscopy revealed a tumorous lesion in the mucosa in the right upper lobe anterior segment. Pathological analysis of a sample led to a diagnosis of bronchioloalveolar carcinoma.

Key words: *Peutz-Jeghers syndrome, bronchioloalveolar carcinoma, lung.*

Özet

Peutz-Jeghers sendromu nadir görülen otozomal dominant geçişli herediter bir hastalıktır. Peutz-Jeghers sendromunda kanser prevalansında artış vardır. Peutz-Jeghers sendromlu 48 yaşında bir erkek hasta bilgisayarlı akciğer tomografisinde bilateral konsolide alanlar ile kliniğimize yatırıldı. Bronkoskopide sağ üst lob anterior segment bronşu içinde tümoral lezyon görüldü. Patolojik tetkikde bronkoalveoler kanser tanısı kondu.

Anahtar Sözcükler: *Peutz-Jeghers sendromu, bronkoalveoler karsinom, akciğer.*

Lung cancer is the leading cause of cancer-related death all over the world. Unfortunately, at first diagnosis, the majority of patients have an advanced stage of the illness. The stage observed upon diagnosis greatly affects survival. Early detection and diagnosis is the key to improving the survival rate of lung cancer patients. In addition, in rare cases, some syndromes may coexist with lung cancer, such as PJS (1,2).

PJS is an autosomal dominant inherited disease characterized by intestinal hamartomatous polyps and mucocutaneous pigmentation associated with a serine-threonine kinase 11 (STK11) mutation (3). In 1921, Peutz (4) described a Dutch family with multiple intestinal hamartomatous polyps and characteristic melanin spots on the skin and mucosa. In 1949 Jeghers et al. (5) described 3 more families with identical lesions seen at the Johns Hopkins Clinic. The original name of Peutz-Jeghers syndrome was first used in 1954 (6).

The incidence of the syndrome is estimated to be 1 in 50,000 to 200,000 (7). Mucocutaneous pigmented lesions have been observed in 95% of patients, which may be the first finding of the syndrome. These lesions may be seen at birth. The classic pigmented lesions may be found on the lips, in the mouth, on the nose, in the perianal area, on the fingers, or in the dorsal and volar regions of the hands and feet. These pigmented macules are probably the result of a melanin increase in the basal cells due to an inflammatory block in the melanin migration (8-10).

The most important clinical problem in patients with PJS is the mechanical complications that arise

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from intestinal polyps. Polyps may be seen in the entire gastrointestinal system. However, they are usually localized in the small intestine and the colon. Other sites include the gall bladder, bronchia, bladder, and the ureter. Gastrointestinal polyps may lead to bleeding, anemia, and abdominal pain.

CASE

A 48-year-old male patient presented at the clinic in August 2017 with the complaints of a cough and white foamy phlegm. A physical examination revealed brown spots in the perioral and perinasal area, and blue mucocutaneous lesions were observed in the mouth (Figure 1). His history included 28 pack-years of smoking. The pigmented lesions on his face and in his mouth were present at birth. He had suffered from abdominal pain since childhood, and had undergone colon polypectomy operations a total of 4 times, with the last operation in 2010, with the diagnosis of Peutz-Jeghers syndrome (PJS). His mother also had pigmented lesions in the perioral area. The patient's daughter had undergone 2 polypectomies, and his son had undergone 3 polypectomy operations with the diagnosis of PJS.

Thorax tomography revealed consolidated areas containing air bronchograms in the right upper lobe posterior and middle lobe lateral segments, and patchy frosted glass areas in both lungs, which was more obvious on the right, a nodular lesion 17x14 mm in size in the left upper lobe, and common micronodules (Figure 2). A positron emission tomography examination revealed a hypermetabolic mass lesion in the right upper lobe posterior segment. An upper abdominal ultrasonography revealed lesions 32x24 mm in size in the lateral segment of the left lobe of the liver and 37x42 mm in the caudate lobe with a relatively smooth contour, ISO-light compared with the liver parenchyma, and compatible with hyperechoic hemangioma. In the contrastive dynamic magnetic resonance imaging (MRI) of the abdomen, 5 or 6 well-circumscribed mass lesions 49x41 mm in size with a lobular contour, of which the largest was located in the caudal lobe and near the inferior vena cava, and which were incompatible with hemangioma, were observed in the liver. No pathology was observed in a brain MRI apart from bilateral maxillary sinus, mucosal thickening, and a retention cyst on the left.

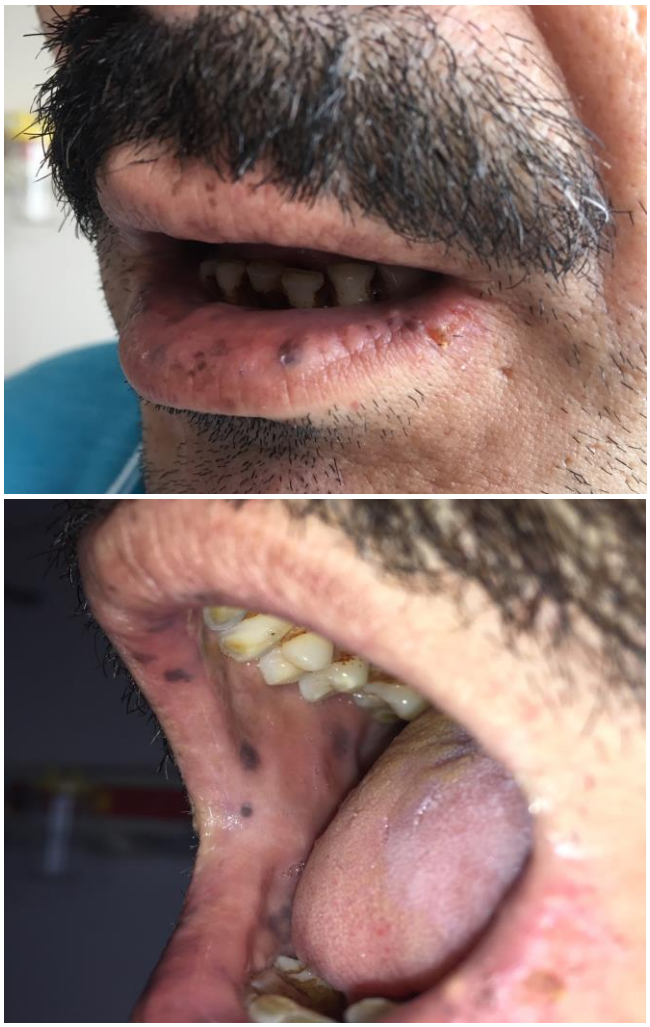

Figure 1: *Perioral and mucocutaneous lesions*

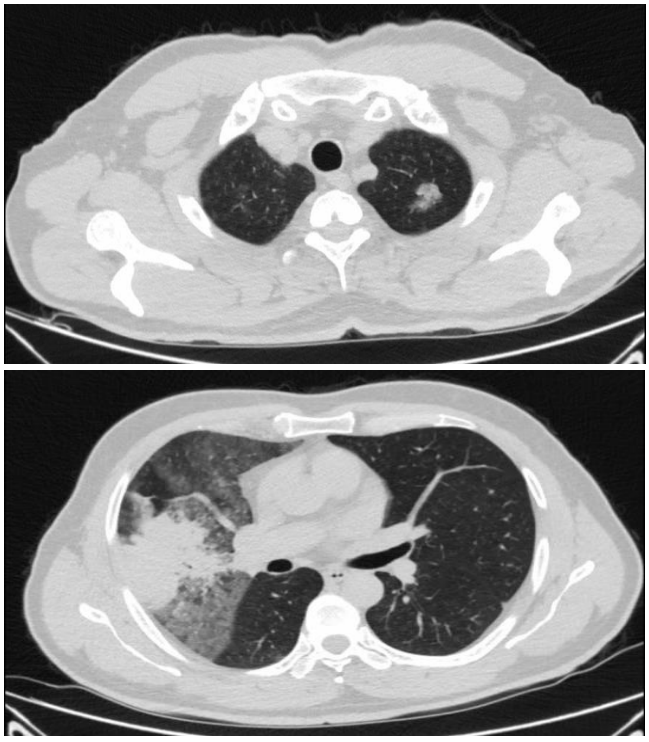

Figure 2: *Thorax computed tomography images*

Bronchoscopy revealed a tumorous lesion in the mucosa in the right upper lobe anterior segment carina separation, and a biopsy was performed. In addition, a transbronchial lung biopsy sample was taken from the middle lobe lateral segment. Bronchioloalveolar lung carcinoma was diagnosed based on the pathological examination (Figure 3).

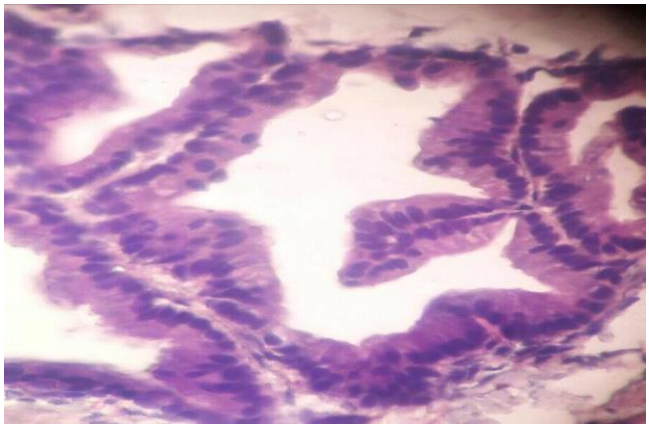

Figure 3: *Bronchioloalveolar lung carcinoma (H&E; original magnification x100)*

DISCUSSION

In this case report, we presented a patient with PJS who developed bronchoalveolar carcinoma, which is a rare and unusual type of lung cancer. To the best of our knowledge, this is the first such case published in our country. A total of 143 patients with PJS had been reported as of 2000. However, only 6 of these patients had lung cancer.

Epidemiological studies show that there is an elevation in the prevalence of cancer as well as non-malignant gonadal tumors in patients with PJS. The risk of cancer is thought to be increased in PJS due to a hamartoma-adenocarcinoma relationship. The presence of adenomatous foci within the polyps of PJS supports this hypothesis. However, cancer is also seen in organs where there are no hamartomas associated with the syndrome (11). It has been suggested that PJS polyps have no potential for malignancy, given the rarity of transformation, and that the polyps are not hamartoma, but only abnormal mucosal prolapse associated with the genetic mutation. (12). Thus, the increased risk of cancer in patients with PJS may be attributed to mucosal instability through the conventional neoplastic pathways.

In a case series, Li et al. (13) and Giardiello et al. (14) reported cancer patients at the rate of 28% and 22% cancer patients were reported in their series. Giardiello et al. (15) carried out a meta-analysis of 210 individuals evaluated in 6 trials. Lung cancer was responsible for 15% of cancer cases. Hearle et al. (16) then published a cohort study that involved 419 individuals with PJS. They analyzed the incidence of cancer and found that the STK11 mutation was present in 297 of the patients, and that different organ cancers developed in 23% of the patients. The risk of developing all types of cancer increased after the age of 50 years. In the series of Mehenni et al. (17) with 149 patients, 31 malignancies were detected in patients with STK11-mutation PJS, and none of those patients had lung cancer.

CONFLICTS OF INTEREST

None declared.

AUTHOR CONTRIBUTIONS

Concept - B.Y., B.T., M.S.; Planning and Design - B.Y., B.T., M.S.; Supervision - B.Y., B.T., M.S.; Funding -; Materials -; Data Collection and/or Processing -; Analysis and/or Interpretation -; Literature Review -; Writing -; Critical Review -

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