Metachronous tumors in a patient with Fanconi's anemia: case report

ABSTRACT

We present the case of a patient who entered early menopause at age 29 and was diagnosed with two primary tumors, bladder cancer at age 30 and esophageal cancer at 36. She has family history of cancer, since her brother had esophageal cancer at age 29. Given her clinical status, we hypothesized Fanconi anemia, which was diagnosed by the diepoxybutane (DEB) test. The literature contains few case reports on the association between Fanconi anemia and esophageal cancer, and only one regarding its association with bladder cancer.

Keywords: Fanconi anemia, esophageal cancer, bladder cancer, case report.

INTRODUCTION

Fanconi anemia (FA) is a rare autosomal recessive genetic disorder that mainly affects young adults. It is characterized by pancytopenia, congenital malformations and endocrinological disorders, and findings of early menopause and infertility are common. Predisposition for the occurrence of hematological and non-hematological malignancies, especially hepatic malignant neoplasia and squamous cell carcinoma (SCC) of the upper digestive tract, upper airway and anogenital region, has also been reported. DNA binding agents, such as diepoxybutane (DEB), have been used as diagnostic tests.

CASE REPORT

The patient was a 37-year-old woman, ex-smoker (15 years smoking 1 pack/day), with a body mass index of 24.6 kg/m² and family history of esophageal neoplasia (brother, at age 29). At the age of 29, she entered menopause and at age 30 was diagnosed with low-grade bladder urothelial cancer, which was treated by local resection. Six years later, she developed a chronic dry cough and CT scans revealed pulmonary thromboembolism, the presence of a
pulmonary nodule measuring 1.1 cm in the middle lobe, left and cervical supraclavicular (smallest diameter 1.5 cm) and mediastinal lymph node enlargement (smallest diameter 2.2 cm). Anatomopathological investigation of the cervical lymphadenomegaly revealed a hidden primary lesion of SCC. Given the symptomatology, the first line of chemotherapy – cisplatin plus gemcitabine – was initiated as a matter of urgency, combined with intense hemoptysis after the first application. The patient was submitted to bronchoscopy and digestive endoscopy, which revealed the presence of an ulcerated lesion in the middle third of the esophagus. A biopsy confirmed invasive esophageal SCC. Mediastinal radiotherapy (dose: 3000Gy) was prescribed based on the marked reduction in tracheal lumen caused by tumoral infiltration of the mucosa. Positron emission computed tomography (PET-CT) revealed disease progression, thickening in the middle third of the esophagus (SUV 7.6); cervical (1 cm; SUV 13.3) and paratracheal lymph node enlargement (0.8 cm; SUV 8.5), nodular pericardial formations (up to 1.9 cm; SUV 19.3), multiple pulmonary nodules (SUV 8.9), hepatic nodules (1.1 cm; SUV 9.1) and lymph node enlargement in the hepatogastric recess (SUV 7.6). A second line of chemotherapy was initiated, carboplatin plus paclitaxel weekly, but the patient presented myelotoxicity after two cycles and required hospitalization for febrile neutropenia. PET-CT showed evidence of esophageal, lymph node, pulmonary and hepatic disease progression, associated with pleural and pericardial effusion, with evolution to cardiac tamponade and the need for drainage. Faced with the disease severity and the drop in treatment performance, the medical team opted for exclusive palliative care. A diagnosis of FA was suspected because of her clinical history of cancer, prior early menopause and family history. On the day of her death, a DEB test was performed that was positive for FA.

DISCUSSION

Reports in the literature regarding the association between Fanconi anemia and esophageal cancer are scarce. The highest incidence occurs in young women not submitted to bone marrow transplantation. Kutler et al., Rosenberg et al.,4 and Rosenberg et al.,9 described a profile of cancer incidence in patients with FA. Relating the cases of esophageal cancer to the number of solid malignancies, they found a ratio of 1/79, 2/18, 2/12, respectively. In a review of the literature, Shimamura and Alter10 found 124 patients who presented associations between FA and a solid malignant neoplasm. Of these, 14 patients had esophageal cancer and only one male patient had bladder cancer. Thus, the patient discussed here is the second to be diagnosed with FA and bladder cancer and the first woman reported with this association.

While following eight patients with FA, Itskoviz et al.11 diagnosed esophageal cancer in two patients, 8 and 18 months after the onset of endoscopic follow-up. Due to the increased risk of developing neoplasms of the upper digestive tract, early and periodic endoscopic follow-up is an important feature of patient care.

CONCLUSION

In the general population, the occurrence of FA is rare and this can cause late diagnoses, inadequate follow-up and a decrease in patient survival. This case shows that in young patients with endocrinological disorders, a diagnosis of malignant neoplasia and with family history of cancer, genetic syndromes like FA should be included in the differential diagnosis. Moreover, early diagnostic tests for correct genetic counseling and regular follow-up of patients and relatives are recommended.

REFERENCES

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Corresponding author:
Luis Eduardo Silva Móz
Avenida Alcântara Machado, 2576, Mooca, São Paulo, CEP 03102-002
(14) 99776-3115
lesmoz@uol.com.br