HEREDITARY HEMORRHAGIC TELANGIECTASIA
MIMICKING METASTASES IN A PATIENT WITH GASTRIC CARCINOMA

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Abstract: Hereditary hemorrhagic telangiectasia (HHT) is associated with arteriovenous malformation in multiple organs. The association of HHT with primary malignancy has rarely been reported. We describe the case of a 68-year-old man with gastric carcinoma who presented with abdominal fullness and cramping pain. Radiographic examination showed multiple pulmonary nodules and an osteolytic cervical spine lesion. The initial diagnosis of gastric cancer with multiple metastases was revised after meticulous imaging studies revealed these lesions to be vascular malformations in the lungs and vertebra. This case demonstrates that HHT may coexist with a primary malignancy and mimic multiple metastases.

Case Report

A 68-year-old male presented with abdominal fullness and cramping pain. Endoscopy showed a circular infiltrative lesion over the anterior lesser curvature side extending to the posterior wall of the low body and antrum, with friable mucosa and no definite ulceration. Bormann’s type 4 gastric carcinoma was suspected and histopathologically proved. Malignant metastases were suspected from roentgenographic findings of multiple round nodules in bilateral lungs (Fig. 1) and an osteolytic lesion in the fourth cervical vertebral body (Fig. 2). The chest roentgenogram revealed faint tubular structures connecting the pulmonary nodules to hilar vessels, which prompted further examination of the chest with CT, confirming that these masses were pulmonary AVMs (Fig. 3). Detailed history taking revealed that the patient had suffered from iron deficiency anemia due to recurrent epistaxis for 20 years.

Hereditary hemorrhagic telangiectasia (HHT), also known as Osler-Weber-Rendu disease, is an autosomal dominant disease characterized by systemic vascular dysplasia. The incidence ranges from 1 or 2 per 100,000 to more than 1 per 10,000 in different regions [1, 2]. Systemic vascular dysplasia in varied forms, from telangiectasis to arteriovenous malformation (AVM), can be found in almost all organ systems. Telangiectases mostly occur on the face, lips and tongue, and in the nasal mucosa. The lung, liver and intestine are the more common sites of AVMs. Central nervous system involvement is less common, and spinal lesions have been sporadically reported [3]. While HHT involves multiple organs simultaneously, AVMs of HHT are frequently asymptomatic.

Although the exact pathogenesis is still unclear, HHT may rarely be associated with malignancies such as hepatocellular carcinoma [4], colon cancer [5, 6], and lymphoproliferative malignancies [7]. In a patient with both malignancy and HHT, the multiple AVMs in multiple organs may mimic malignant metastases if HHT has not been diagnosed. The meticulous study of diagnostic images, using tools such as computerized tomography (CT) and magnetic resonance imaging (MRI), is important for correct diagnosis and treatment. We report a case of gastric carcinoma and HHT with multiple pulmonary and cervical vertebral masses mimicking malignant metastases.

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first episode had occurred when he was 30+ years old, and he had received multiple blood transfusions. His father and son also had a history of recurrent epistaxis. Mucocutaneous telangiectasia was not found. The diagnosis of HHT was made based on the findings of recurrent epistaxis, pulmonary AVM, and family history of epistaxis.

MRI was performed to determine the characteristics of the osteolytic lesion of the cervical spine. Conventional imaging revealed a mass with signal void in the fourth cervical vertebra. MR angiography showed a flow signal inside the mass (Fig. 4). The coexistence of gastric carcinoma with HHT was considered. Exploratory laparotomy with radical subtotal gastrectomy was performed, revealing stage IIIa gastric cancer, T3N1M0. Chemotherapy with 5-fluorouracil and leucovorin was started after surgery. The patient died of abdominal carcinomatosis 2.5 years later.

**Fig. 1.** Chest roentgenograms showing multiple well-defined nodules in the bilateral lungs with connecting vascular shadows (arrows). A) Posteroanterior projection; B) lateral projection.

**Fig. 2.** Cervical spine roentgenogram showing an osteolytic lesion in the fourth vertebra (arrows).

**Fig. 3.** Computerized tomography (CT) of the chest showing multiple well-defined nodules with engorged supply and drainage vessels (arrows). A) Contrast-enhanced CT; B) lung window.
Discussion

While the precise pathogenesis is still unknown, HHT is considered to be a congenital malformation of the total vascular system [2], caused by a benign proliferation of tissue in the embryonic stage similar to that of hamartoma. Histologically, the telangiectasia consists of dilated, thin-walled, small arteries, capillaries and venules [2]. Association of HHT with malignancy has been reported occasionally, with histopathogenesis in the angiodysplastic change in involved organs [8]. In our patient, we did not find angiodysplasia grossly in the nontumorous portion nor microscopically in the tumorous portion of the gastric mucosa. The findings suggested that the HHT and gastric cancer were not related.

The spread of gastric carcinoma may occur by direct extension, and via lymphatic, hematogenous and peritoneal routes. The lung is involved in about 20 to 30% of cases and the bone is involved in about 10% [9]. Parenchymal nodules are the most common manifestation of lung metastasis from gastric carcinoma. The thoracolumbar region is the most common site of spinal metastasis from gastric carcinoma, and purely osteolytic lesions are more typical than either mixed osteolytic-osteosclerotic or purely osteosclerotic lesions. The roentgenographic findings in our patient are characteristic of lung and spinal metastases from gastric carcinoma.

Pulmonary AVM, usually multiple, is one of the most common manifestations of HHT. The proportion of asymptomatic patients varied from 13 to 56% in different series reviewed by Burke et al [10]. Suspicion of a vascular lesion should be raised if tubular feeding and draining vessels can be identified roentgenographically. Remy et al reported a CT detection rate of 98.2% for pulmonary AVMs [11]. CT, due to its noninvasiveness and detailed anatomic information, is now the diagnostic modality of choice for pulmonary vascular diseases including pulmonary thromboembolism and AVMs.

The most interesting as well as misleading feature in the clinical presentation of this case was the AVM in the cervical vertebra. The patient’s complaint of nuchal soreness led to suspicion of vertebral metastasis. Spinal involvement in HHT is a rare occurrence. About 8 to 12% of HHT patients have neurologic symptoms, mostly attributed to pulmonary AVMs with right to left shunt resulting in either hypoxemia or embolic episodes due to bypassing of the pulmonary filtering system [2]. Only 8% of neurologically symptomatic cases are due to spinal vascular malformations [3]. MRI, with its advantages of noninvasiveness, versatile tissue contrast, and multiplanar imaging, is the modality of choice for imaging the spine, especially the cervical spine. Furthermore, flow phenomena can be detected intrinsically by variable MRI sequences and are useful in interpreting MR angiography.

This case demonstrates the importance of awareness of HHT in the management of patients presenting with a primary malignancy and multiple masses. Familiarity with imaging modalities is also important to confirm suspicion of HHT, particularly in patients who have a recent diagnosis of malignancy but no prior evidence of HHT.

References


