

## Physical Examination Trumps Mediastinoscopy in Diagnosing Maffucci Syndrome: A Rare Cause of Mediastinal Mass

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WE report a patient who presented with a large thoracic mass but who avoided a potentially catastrophic procedure when the anesthesiologist suggested a simple skin biopsy as an alternative to biopsy *via* a mediastinoscope. Maffucci syndrome is a rare cause of mediastinal mass, but the principles at play in this case report are common and important and highlight the role of the anesthesiologist as consultant.

### Case Report

A 50-yr-old male with medical history significant for chondrosarcoma of the left tibia, Kaposi's sarcoma (non-human immunodeficiency virus), neurofibromatosis, hypertension, chronic atrial fibrillation, and gastroesophageal reflux disease presented to the emergency room with a chief complaint of recent-onset chest palpitations that seemed to be brought on by eating. His past surgical history was significant only for a left total knee arthroplasty in conjunction with the resection of his chondrosarcoma in 1975. Physical exam was significant for well-tolerated tachycardia of 160–170 beats/min, an irregularly irregular rhythm with a grade 2/6 systolic murmur best heard at the apex, and multiple soft, bluish, raised subcutaneous nodules on his trunk, hands, and feet. Laboratory results were significant for mild anemia and an electrocardiogram indicating atrial fibrillation with a ventricular response of 170. The patient was admitted to the medical intensive care unit for heart rate control and to rule out myocardial infarction.

A myocardial infarction was ruled out, and his atrial fibrillation was controlled. However, an anteroposterior chest x-ray revealed a widened mediastinum, and the trans-thoracic echocardiogram performed to exclude an intracardiac thrombus before cardioversion showed a mediastinal mass. This was confirmed by computerized tomography to be a large, heterogenous mass between the right ventricle and the airways (fig. 1). The medical oncologist considered a differential diagnosis of Kaposi's sarcoma, recurrent sarcoma, or a third primary tumor. Urine vanillylmandelic acid and metanephrines were negative. Follow-up, outpatient magnetic resonance imaging indicated that the mass was in close proximity to the bronchi and trachea, suggesting that tissue could be obtained by mediastinoscopy. The cardiothoracic surgeons felt that the lesion was not resectable, but a tissue diagnosis might be possible *via* mediastinoscopy.

During preoperative evaluation for mediastinoscopy, the anesthesiologist noted the presence of multiple subcutaneous nodules consistent with neurofibromatosis noted in patient's history. The lesions were compressible and shrunk with elevation of the hands above the heart. Additional inquiry of the patient revealed that, despite his presumptive diagnosis of neurofibromatosis, the subcutaneous nodules had never been biopsied. The anesthesiologist theorized that the lesions on the patient's hands could be of the same origin as that in his mediastinum. After discussion with the cardiothoracic surgeon and medical team, the patient underwent the much less invasive diagnostic biopsy of two cutaneous nodules instead of biopsying the large mediastinal tumor.

The tissue revealed cavernous hemangioma with calcifications and papillary endothelial hyperplasia adjacent to the hemangioma. Under high power magnification, pericytic proliferation was seen and immunohistochemistry revealed positive staining for CD34+, an endothelial marker. The diagnosis was papillary hemangioendothelioma. It was assumed that the mediastinal mass was also a hemangioma. A literature search crossing the terms "hemangiomas" and a "chondrosarcoma" yielded the diagnosis of Maffucci syndrome.

One month later, the patient returned to the emergency room complaining of gross hemoptysis. The patient was admitted to the medical intensive care unit and an emergency bronchoscopy revealed bright red blood rising from the right mainstem bronchus. On the basis of previous findings and the diagnosis of Maffucci syndrome, it was felt that the bleeding arose from the mediastinal lesion, and this was confirmed by an angiogram that showed a tumor blush when the right bronchial artery was injected (fig. 2). Branches of the right bronchial artery that fed the mediastinal lesion were successfully embolized and stopped the hemoptysis. The patient recovered without incident.

### Discussion

Maffucci Syndrome is a rare congenital disorder of mesodermal dysplasia consisting of enchondromatosis and subcutaneous hemangiomas. Enchondromatosis alone (Ollier's disease) comprises multiple benign tumors of mature hyaline cartilage (enchondromas) in the metaphysis of long bones and digits, frequently resulting in limb deformities, short stature, or scoliosis.<sup>1</sup> Benign lesions have an irregular distribution but frequently appear on radiography to have well-defined boundaries. Tumors have a high rate of malignant transformation; for example, chondrosarcomas, the most common type of enchondroma, occur in 30% of reported Maffucci cases.<sup>2</sup> The second criteria for Maffucci syndrome is the evidence of vascular malformations or hemangiomas. These tumors appear as soft blue subcutaneous nodules and are frequently located in the distal digits. The lesions often arise from venous or lymphatic channels and can spread to form highly vascular mass in the trunk, such as the mediastinal mass seen in the patient described above.<sup>3</sup>

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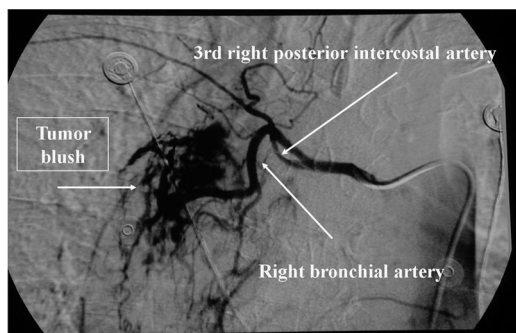
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**Fig. 1.** Representative image from the infusion computerized tomography shows a large posterior mediastinal, inhomogeneous mass located between the heart and the descending aorta.

Although the clinical classification of Maffucci syndrome is well defined, physical manifestation of the disease is often heterogeneous. Symptoms are not present at birth but generally occur in early childhood with several pleiotropic phenotypes.<sup>3</sup> Skeletal deformations associated with enchondromas are variable, affecting both long and flat bones,<sup>4</sup> and malignant transformation to chondrosarcoma is common. Furthermore, a broad spectrum of other cellular dysplasias has been reported, including ovarian, pancreatic, parathyroid, and pituitary tumors.<sup>5-7</sup> A recent study suggests a receptor mutation for the parathyroid hormone, and related proteins may play a role in the broad spectrum of associated phenotypes.<sup>8</sup> However, another study failed to confirm this finding, suggesting that multiple mechanisms are involved in the pathogenesis.<sup>9</sup> Similarly, vascular lesions, although easily identified by small blue subcutaneous nodules, may appear in various other sites. Large hemangiomas have been sited within the brain, orbit, gastrointestinal tract, and abdominal wall.<sup>10-15</sup> Hemangiomas can also undergo malignant transformation to become hemangiosarcoma, lymphangiosarcoma, or hemangioendothelioma, as seen in the patient described above.<sup>16</sup> Since the first report by Maffucci in 1881, only about 200 cases have been reported worldwide, including two cases of thoracic vascular tumor similar to that observed in our patient.



**Fig. 2.** From this film, it is apparent that the branch of the bronchial artery will be reachable for embolization.

The diagnosis and treatment of this rare thoracic vascular tumor provides a valuable lesson in the practice of medicine in general and anesthesiology specifically. The standard procedure to determine the diagnosis of the tumor was biopsy, a procedure which, given the vascular origin of its development, would likely have resulted in severe hemorrhage in a site where bleeding would be difficult to control. This potentially fatal complication was avoided when the anesthesiologist observed that the subcutaneous nodules could provide an alternative site for biopsy. Although often overlooked, symptoms or physical signs not apparently related to the presumed diagnosis can provide important diagnostic clues. Recognizing these details requires careful observation and clinical suspicion, especially when the diagnosis is not known. Careful clinical judgment can thereby improve patient safety by reducing the likelihood of an unwarranted and potentially dangerous procedure. This report illustrates the importance of careful observation and clinical suspicion that lie at the foundation of the practice of medicine. Furthermore, we highlight the valuable role in which anesthesiologist act as consultants and patient advocates, thereby contributing not only to patient safety but to overall clinical diagnosis and patient management.

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