DIAGNOSING A FREAK OF NATURE

By Vanessa Jeanne Gray

Our patient presented to the emergency department with tearing chest pain radiating to his back. His presenting symptoms, coupled with his history of hypertension and a chest x-ray that showed significant mediastinal widening, made us sure he was experiencing an aortic dissection. But our diagnostic CT scan showed that we were in for a much bigger surprise.

More than 250 years ago in London, Dr. David Bayford, an apprentice surgeon, attended the autopsy of a woman who had died of dysphagia-induced starvation. At first, no cause for her dysphagia could be discovered. However, upon further investigation, Dr. Bayford found an aberrant right subclavian artery that compressed upon, and essentially strangled, her esophagus. Impressed by the appearance of the anomaly he saw, Dr. Bayford described the woman’s condition as dysphagia lusoria from the Latin term lusus naturae, meaning “freak of nature.”

In 1936 in Berlin, Dr. Burckhard Kommerell was one of the first physicians to clinically diagnose the same congenital anomaly, which previously had only been discovered during autopsy. He did this, surprisingly, in a patient who did not present with dysphagia.

A diagnostic radiologist, Dr. Kommerell was viewing a barium swallow of a 65-year-old man presumed to have stomach cancer. In the sagittal view, the barium swallow demonstrated a delay of the contrast material past the aortic knob. In a more oblique view, the image showed compression of the esophagus at the same location by a pulsating mass. Dr. Kommerell knew that what he was seeing was more than just the aberrant right subclavian artery that had previously been described by Dr. Bayford. Dr. Kommerell identified the pulsating mass as an aortic diverticulum, a rare complication of an aberrant right subclavian artery and an anomaly that has since carried his name.

Caused by the incomplete embryonic development of the right, fourth aortic arch, an aberrant right subclavian artery (ARSA) is found in approximately 1% of the total population. Normally, the right subclavian branches off the brachiocephalic artery and is the blood supply to the right upper extremity. An ARSA, however, has an anomalous origin directly off the aortic arch as a fourth branch, just lateral to the left subclavian. In 80% of cases, it will pass posterior to the esophagus as it continues on its normal course to provide the blood supply to the right arm. As in Dr. Bayford’s autopsy case, the ARSA can compress the esophagus as it passes posteriorly, leading to the most common complaint of dysphagia. Other possible complications of an ARSA include atherosclerosis, stenosis, aneurysms and dissections.

A diverticulum of Kommerell (DOK) aneurysm is a dilatation of the proximal portion of the aberrant right subclavian as it comes directly off the aortic arch. Found in only 0.5% of the population, a DOK aneurysm is potentially fatal if not diagnosed promptly and accurately. Like the ARSA, it too can cause symptoms of dysphagia in addition to other non-specific complaints such as cough, shortness of breath and, as we found in our patient, chest pain.

Thanks to current technology, we no longer have to wait for an autopsy to make the diagnosis of an aberrant subclavian artery complicated by a diverticulum of Kommerell aneurysm. While an aortogram is the gold standard for such a diagnosis, our patient was able to be diagnosed by a CT scan ordered to rule out what we initially thought was a sole aortic dissection. From the CT scan, we discovered that our patient not only suffered from a combined ARSA and DOK aneurysm, but that he did in fact have a Type B dissection as well.

Having any one of these three entities is very rare, but to have them all concurrently is exceedingly unusual. A 2005 study in Cardiovascular Interventional Radiology found that of the 2,400 thoracic aortograms performed at a Level 1 trauma center over 12.5 years, only nineteen showed an aberrant right subclavian artery. Of those nineteen, seven had an associated diverticulum of Kommerell and, of those seven, only one also had a Type B dissection.

Thus, while it is remarkably rare to find an aberrant right subclavian artery, a diverticulum of Kommerell aneurysm and a Type B dissection occurring simultaneously, our experience shows the importance of using a systematic approach and having a broad differential diagnosis when working up a patient with chest pain in order to prevent potentially fatal complications.

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