A 28-year-old, previously healthy man visited a gastroenterologist with symptoms that included regurgitation of foods and liquids when in the supine position or while bending over. He also had paroxysms of coughing and an unintentional weight loss of 20 lbs (9.1 kg). These symptoms developed during a 6-month period and did not respond to dietary modifications, sleeping with the head of the bed elevated, or treatment with proton-pump inhibitors. The patient denied frank heartburn, dysphagia, chest pain, nausea, and loss of appetite. He had no history of tobacco or alcohol use. Results of his physical examination were normal. A radiograph image was obtained (Figure 1).

What Is the Diagnosis?

A. Hiatal hernia
B. Zenker’s diverticulum
C. Achalasia
D. Esophageal cancer

Figure 1.
Achalasia is a functional disorder of esophageal motility. With an annual incidence of only 0.6 per 100,000 individuals, it usually affects patients between the ages of 20 and 40 years. The defining abnormalities are absence of esophageal peristalsis and failure of the lower esophageal sphincter (LES) to relax completely on swallowing, thus leading to the chronic, irreversible condition of a dilated esophagus. Achalasia is currently thought to result from neurogenic defects in the esophageal myenteric plexus, which, in turn, may be caused by autoimmune disease, infection, or primary neuron degeneration.

Recognition of the typical symptoms of achalasia guides the appropriate patient evaluation. Approximately 75% of patients relate a history of regurgitation, dysphagia, and weight loss, while another 50% may experience heartburn and chest pain. These symptoms should warrant the performance of a barium swallow, which usually reveals the characteristic features of “bird’s-beak” narrowing of the distal esophagus and proximal esophageal dilation (Figure 1). A complete assessment, however, requires esophageal and gastric endoscopy and esophageal manometry studies. Abnormal parameters confirmed by manometry include a hypertensive (>35 mm Hg) nonrelaxing LES, increased esophageal pressure, and decreased peristalsis. If esophageal motility is found to be normal, a search for extraesophageal pathological conditions mimicking achalasia (e.g., pseudoachalasia) is necessary. A computed tomography scan of the chest and abdomen is helpful to detect a regional mass, cancer, or an aortic aneurysm.

The goal of treating achalasia is to improve esophageal emptying. By the time a patient obtains surgical referral, several treatment strategies may have been tried, including calcium channel blockers, pneumatic and bougienage dilation, and injection of botulinum toxin. All of these treatment plans are directed at disrupting or paralyzing the hypertensive LES. Surgical transection of the LES fibers (the Heller cardiomyotomy procedure) has been accomplished through abdominal, thoracic, and laparoscopic approaches. Surgery is the appropriate treatment for the following 4 types of patients: (1) young patients, in whom pneumatic dilation has been shown to have poor long-term results; (2) those with recurrent symptoms after undergoing nonsurgical interventions; (3) patients at high risk for complications from pneumatic dilation, such as those with a tortuous esophagus or esophageal diverticulitis; and (4) patients who request surgery as the initial treatment. Results from surgery are very favorable, with low esophageal perforation rates (<1%) and durable long-term symptom relief. Because cardiomyotomy lowers the LES resting pressures to levels that allow gastric reflux, a partial fundoplication is a recommended component of the operation. The laparoscopic approach is being increasingly applied toward effective treatment of this condition (Figure 2).

References