

ESOPHAGEAL LEIOMYOSARCOMA PRESENTING WITH RECURRENT LARYNGEAL NERVE PALSY AND GREAT VESSEL ENCASEMENT: A CASE REPORT

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Received date: 20 May 2024 Revised date: 18 June 2024

Acceptance date: 07 July 2025

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ABSTRACT

Background: Esophageal leiomyosarcoma is an extremely rare malignancy of the esophagus ($\leq 0.5\%$ of esophageal tumors) and typically presents with dysphagia and weight loss in middle-aged or older patients. Hoarseness of voice due to recurrent laryngeal nerve (RLN) palsy is an exceedingly uncommon initial presentation. We report a case of esophageal leiomyosarcoma in a 55-year-old man who presented with isolated hoarseness, posing a diagnostic challenge due to a normal endoscopic biopsy. **Case Presentation:** A 55-year-old male heavy smoker presented with severe progressive hoarseness. Laryngoscopy confirmed left vocal cord paralysis. Contrast-enhanced CT of the neck and chest revealed a 3.5 cm necrotic soft-tissue mass in the left paraesophageal region encasing the aortic arch and left subclavian artery, with medial displacement of the left cricoarytenoid cartilage and atrophy of left laryngeal muscles consistent with left RLN palsy. Despite these findings, upper endoscopy showed no intraluminal lesion and random esophageal biopsies were normal. Given high suspicion for a submucosal tumor, the patient underwent surgical resection. **Outcome:** Histopathology of the resected esophagus revealed a spindle cell sarcoma consistent with leiomyosarcoma. The resection margins were clear, and no lymph node metastases were identified. The patient had an uneventful recovery. At 3-year follow-up, he remains free of disease with no recurrence and has experienced substantial improvement in his voice. **Conclusion:** This case highlights an unusual presentation of esophageal leiomyosarcoma causing vocal cord paralysis by invading the left RLN. It underscores the importance of pursuing definitive diagnosis in the face of negative endoscopic biopsies when clinical and radiologic suspicion for a submucosal esophageal tumor is high. Early surgical intervention can be both diagnostic and curative in such cases, and complete resection offers the best chance for long-term survival.

INTRODUCTION

Esophageal leiomyosarcoma is a rare malignant tumor arising from smooth muscle in the esophageal wall, accounting for only about 0.1–0.5% of all esophageal malignancies. Fewer than a few hundred cases have been reported in the literature to date. Patients are usually middle-aged or elderly, and common symptoms at presentation include progressive dysphagia (in ~78% of cases) along with weight loss (in ~54%), chest pain (36%), or regurgitation. In contrast, presentations without dysphagia are extremely rare. Hoarseness of voice due to left recurrent laryngeal nerve palsy is an unusual initial manifestation of any esophageal tumor;

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when present, it often indicates an advanced lesion in the upper chest causing Ortner's syndrome (cardiovocal syndrome). We describe a case of esophageal leiomyosarcoma that uniquely presented with isolated hoarseness from RLN involvement. This case posed significant diagnostic challenges, as initial endoscopic biopsy was false-negative – a known pitfall for submucosal esophageal tumors. We discuss the diagnostic approach, imaging and pathology findings, surgical management, and review the relevant literature on esophageal leiomyosarcoma, with particular focus on cases with similar vascular and neural involvement and the importance of aggressive management despite negative biopsies.

CASE PRESENTATION

A 55-year-old man with a history of heavy smoking (40 pack-years) presented to the otolaryngology clinic with a 3-month history of progressive hoarseness of voice. He denied dysphagia, odynophagia, weight loss, or chronic cough. Physical examination revealed a breathy, weak voice. There were no enlarged cervical lymph nodes or other remarkable findings in the head and neck exam. Flexible laryngoscopy demonstrated left vocal cord paralysis in the paramedian position. The rest of the neurological examination was unremarkable. Given the isolated left vocal cord palsy in a long-term smoker, a workup for an apical chest mass or mediastinal pathology compressing the left recurrent laryngeal nerve was initiated.

Investigations

Imaging: A contrast-enhanced CT scan of the neck and thorax was performed to evaluate the cause of the vocal cord palsy. The scan revealed evidence of chronic emphysema in the lungs, but more importantly identified a necrotic soft-tissue mass in the left paratracheal/paraesophageal region at the level of the aortic arch. The mass measured approximately 3.1×3.5 cm and was seen encasing the aortic arch and the proximal left subclavian artery, and abutting the left common carotid artery. There was loss of the fat plane between the mass and the esophagus, suggesting an esophageal origin of the tumor. No definitive intraluminal component was observed on CT, but prominent necrosis was present within the mass. The mass extended into the aortopulmonary window region. Corresponding to this, the CT also showed objective signs of left vocal cord paralysis: there was medial rotation of the left arytenoid (cricoarytenoid) cartilage with atrophy of the left thyroarytenoid muscle and ipsilateral dilatation of the laryngeal ventricle. These findings indicated longstanding left recurrent laryngeal nerve involvement by the tumor. No gross pulmonary lesion was seen aside from centrilobular and paraseptal emphysema with apical bullae, consistent with the patient's smoking history. There were no enlarged distant lymph nodes or obvious distant metastases on imaging.

Endoscopy: An upper gastrointestinal endoscopy was carried out to evaluate the esophageal lumen. The esophageal mucosa appeared generally normal, with no visible mass or ulceration internally. Multiple endoscopic biopsies of the esophageal mucosa were obtained at the level correlating with the imaging findings. Histopathological examination of these initial biopsies was normal, showing only benign squamous mucosa with no evidence of malignancy. This result suggested that the lesion was likely submucosal or extrinsic to the esophagus, such that the mucosa remained uninvolved.

Differential Diagnosis of the mass: Based on the CT findings of a necrotic left paraesophageal mass causing vocal cord palsy, the leading consideration was an esophageal malignancy, particularly a sarcomatous tumor arising from the esophageal wall (given the submucosal

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nature and necrosis). The radiologist's impression favored an esophageal leiomyosarcoma involving the left recurrent laryngeal nerve as the closest diagnosis. Other differential considerations included a primary bronchogenic carcinoma of the left lung apex or mediastinum with metastatic lymphadenopathy causing RLN compression (especially small-cell carcinoma or squamous cell carcinoma, given the patient's heavy smoking history). However, the absence of a lung mass on CT and the localization of the tumor to the esophageal region made a primary esophageal lesion more likely. An esophageal gastrointestinal stromal tumor (GIST) was also considered in the differential, given that GISTs are mesenchymal tumors that can be submucosal; however, esophageal GISTs are extremely rare. Other possibilities, such as a metastatic lymph node or a mediastinal neoplasm (lymphoma, nerve sheath tumor), were less likely but were kept in mind. Overall, the constellation of imaging findings pointed toward a primary esophageal sarcoma (such as leiomyosarcoma) as the most likely diagnosis, despite the negative endoscopic biopsy.

Treatment

Given the high suspicion for a submucosal esophageal malignancy causing compressive symptoms, a decision was made to proceed with surgical exploration and resection both for definitive diagnosis and treatment. The patient underwent a surgical esophagectomy with en bloc resection of the tumor. A left posterolateral thoracotomy approach was utilized to access the upper thoracic esophagus and mediastinum (containing the tumor encasing the great vessels). Intraoperatively, the tumor was found arising from the esophageal wall just below the thoracic inlet, extending into the mediastinum. It was firmly adherent to surrounding structures, including the adventitia of the aortic arch and left subclavian artery, as visualized on imaging. Careful dissection allowed separation of the mass from the vascular structures without the need for vascular resection, although the left recurrent laryngeal nerve, which was already non-functional preoperatively, appeared to be infiltrated by tumor and had to be sacrificed during resection. The esophageal segment containing the tumor was removed in its entirety. Reconstruction was achieved via a cervical esophagogastric anastomosis after gastric pull-up (as is standard after esophagectomy). The surgery was technically challenging due to the tumor's location and its encasement of vital structures, but complete gross resection (R0) was accomplished.

Histopathological examination of the resected specimen confirmed the diagnosis of leiomyosarcoma of the esophagus. The tumor was a high-grade spindle cell sarcoma arising from the muscularis propria of the esophagus. Microscopically, it showed intersecting fascicles of atypical spindle cells with elongated "cigar-shaped" nuclei and frequent mitoses. There was also central necrosis consistent with the radiologic findings. Immunohistochemistry demonstrated strong positivity for smooth muscle actin and desmin, confirming smooth muscle lineage, while markers for gastrointestinal stromal tumor (KIT [CD117] and DOG1) were negative. These findings ruled out a GIST and solidified the diagnosis of leiomyosarcoma. No histological evidence of an epithelial (carcinomatous) component was seen. The tumor appeared to focally invade beyond the esophageal wall and involve surrounding soft tissue, including the area of the left vagus nerve/RLN, but there was no invasion into the lumen or mucosa of the esophagus. All resection margins were clear of tumor. No metastasis was found in the resected regional lymph nodes (0/8 lymph nodes involved).

No adjuvant therapy (chemotherapy or radiotherapy) was administered postoperatively. The rationale was that, in leiomyosarcoma of the esophagus, surgical resection is considered the optimal and potentially curative treatment, and the role of adjuvant therapy is not well established. Given the complete (R0) resection and absence of nodal disease, a surveillance strategy was adopted.

Outcome and Follow-Up

The patient's postoperative course was smooth. He had an uneventful recovery from the esophagectomy, with the anastomosis healing well and no major complications. His preoperative hoarseness persisted in the immediate postoperative period due to the left vocal cord paralysis; however, the patient underwent voice therapy and subsequently a medialization procedure of the left vocal cord to improve glottic closure. This resulted in significant improvement in his voice quality over time. By the 6-month follow-up, he was able to speak with a stronger voice, and he had adapted well, with no aspiration symptoms.

At routine follow-up evaluations, including clinical exams and surveillance imaging (annual CT scans of the chest and neck), there has been no evidence of tumor recurrence. At 3 years after surgery, the patient remains in complete remission and is leading a normal life. He is free of dysphagia or any new symptoms. His initial presenting symptom of hoarseness has greatly improved, and he reports no significant limitations in daily activities. The patient continues to be monitored yearly, given that late recurrences of leiomyosarcoma have been reported in some cases. We obtained written informed consent from the patient to publish the details of his case.



Figure 1: CT Neck Showing Anteromedial Rotation of the Cricothyroid Cartilage

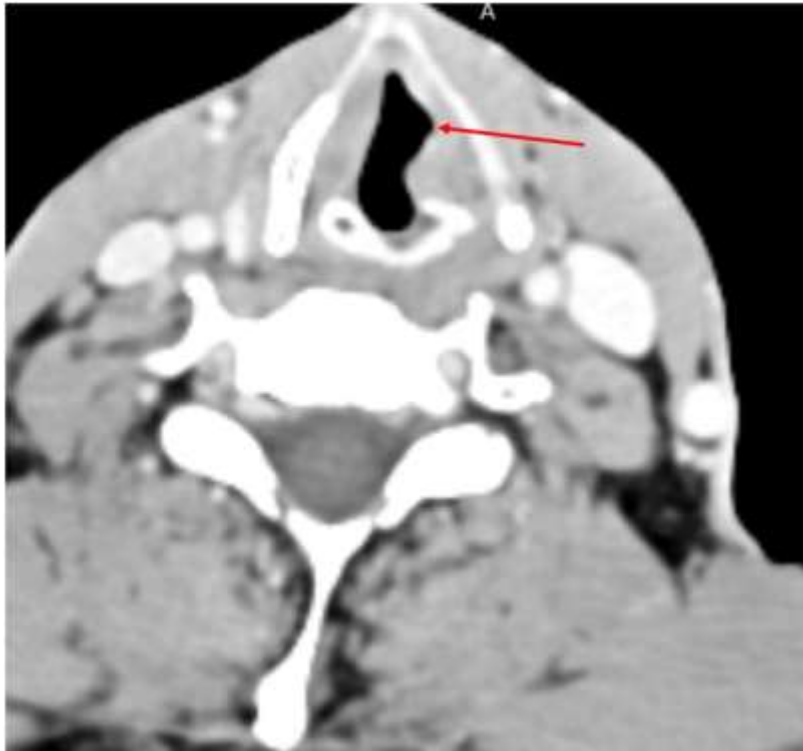


Figure 2: CT Neck Showing Post Contrast Venous Phase Images Dilatation of the Left Lateral Ventricle and Atrophy of the Left Thyroarytenoid.

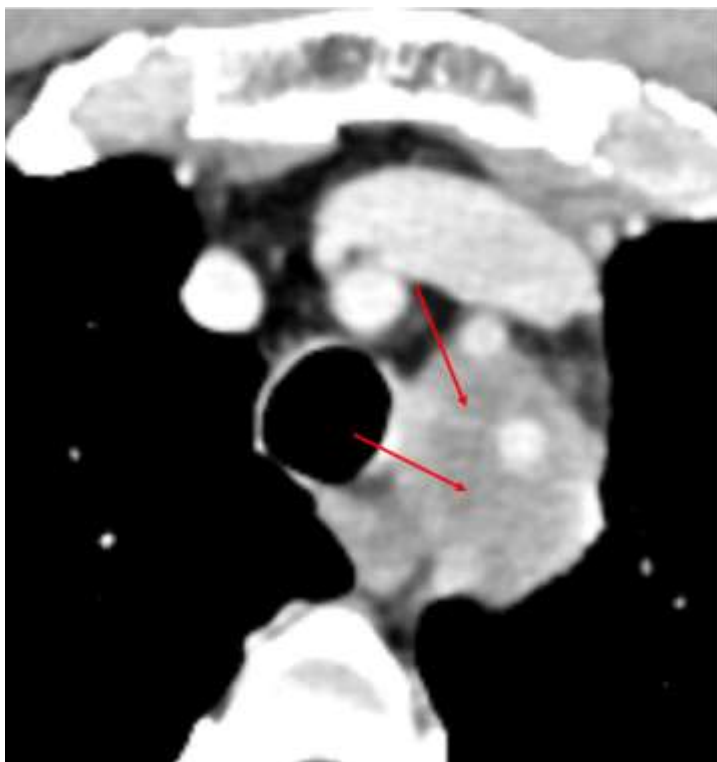


Figure 3: Ct Neck Post Contrast Images Showing Encasement of the Left Subclavian Artery and Left Common Carotid Artery Seen

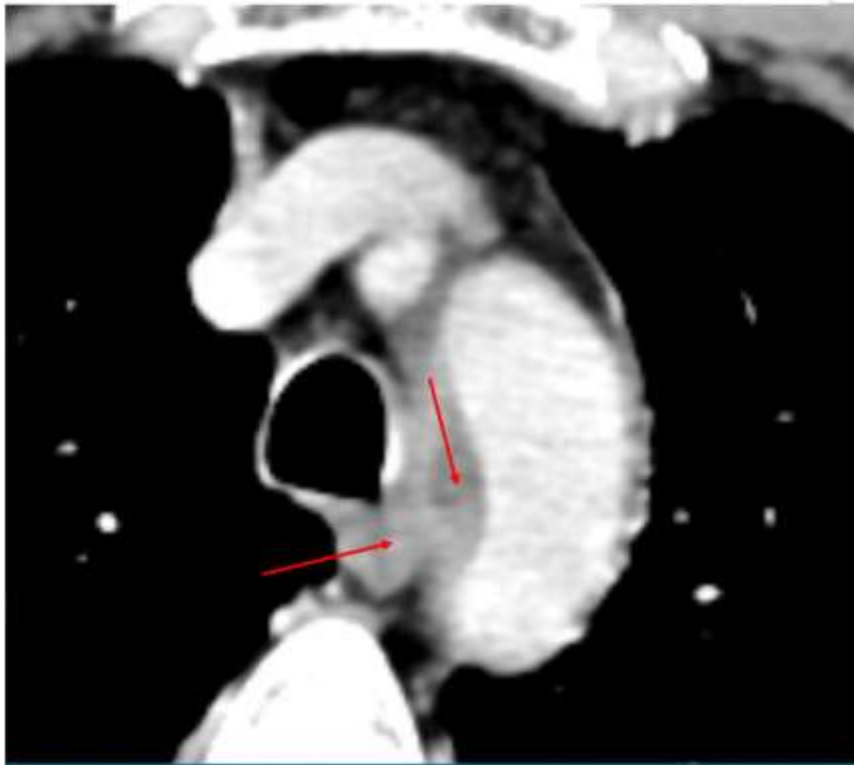


Figure 4: CT Neck Post Contrast at the Aortic Arch Level Showing the Encasement of the Aortic Arch.



Figure 5: Visualized CT Chest Showing Regions of Centrilobular Emphysema and Bulla and Paraseptal Emphysema and Bulla.

DISCUSSION

Overview and rarity: Leiomyosarcoma of the esophagus is an exceedingly rare entity. It is the most common type of esophageal sarcoma, yet esophageal sarcomas themselves comprise well under 1% of esophageal tumors. Since the first description in 1902, only approximately 150–200 cases have been reported in the literature. Patients typically present in the fifth to seventh decade of life, similar to our 55-year-old patient. Unlike the far more prevalent esophageal squamous cell carcinomas and adenocarcinomas, which arise from the mucosa, leiomyosarcomas originate in the muscular layer and often grow in an exophytic or intramural fashion. As a result, they may attain large size before causing lumen obstruction.

Clinical presentation and unusual features: The majority of esophageal leiomyosarcomas present with dysphagia (difficulty swallowing) as the dominant symptom, often accompanied by weight loss or retrosternal pain. Our case is unusual in that the patient's initial and sole presenting complaint was hoarseness of voice, caused by left recurrent laryngeal nerve (RLN) palsy. Hoarseness in esophageal cancer (so-called Ortner's syndrome or cardiovocal syndrome) generally indicates a large upper-esophageal or mediastinal tumor invading or compressing the left RLN where it loops under the aortic arch. In esophageal squamous carcinoma, vocal cord paralysis is recognized as a sign of advanced disease (T4 stage, due to mediastinal invasion). However, it is extremely rare for a primary esophageal leiomyosarcoma to present with RLN palsy, especially in the absence of dysphagia. A review of published cases found only a few reports of esophageal leiomyosarcomas with similar nerve involvement. For example, Jutley et al. reported a leiomyosarcoma discovered incidentally in a patient evaluated for chronic cough, with no dysphagia noted. Our case reinforces that an esophageal mass should be included in the differential diagnosis of isolated left vocal cord paralysis, even if classic esophageal symptoms are lacking.

Additionally, the CT in our case showed the tumor encasing major vascular structures (the aortic arch and left subclavian artery). Leiomyosarcomas are often large and can distort or displace adjacent structures, but they are classically described as having "limited penetration" into surrounding tissues. In other words, they tend to push adjacent organs away rather than widely infiltrating them, which sometimes allows resection even when vital structures are involved. In our patient, despite radiologic encasement of the great vessels, the tumor could be dissected off the vessel walls, suggesting it was abutting and compressing them but not grossly invading the arterial adventitia. This feature is consistent with many sarcomas: they can grow big and wrap around structures without the early micrometastatic spread characteristic of carcinomas. Nonetheless, encasement of the aortic arch by an esophageal tumor is exceptionally rare, and to our knowledge only isolated cases of leiomyosarcomas with such extensive mediastinal involvement have been documented. This underscores that radical surgery can be considered in select cases, even if imaging shows apparent encasement, provided that skilled cardiothoracic surgical support is available.

Diagnostic challenges: The diagnosis of esophageal leiomyosarcoma can be challenging and often requires a high index of suspicion. Endoscopic evaluation is important, but as this case illustrates, mucosal biopsies can be falsely negative. Because leiomyosarcomas arise from the deeper layers of the esophageal wall and often have an intact mucosal surface, routine endoscopic biopsy (which samples only the mucosa) may miss the tumor. In our case, despite a substantial tumor, the overlying mucosa was normal endoscopically and histologically. This limitation of endoscopy is well recognized: one study noted that endoscopic biopsies have a high false-negative rate for submucosal esophageal tumors when the mucosa is uninvolved. Endoscopic ultrasound (EUS) can be extremely helpful in such scenarios. EUS can visualize

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intramural lesions and guide fine-needle aspiration or core biopsy of submucosal masses, improving diagnostic yield. In retrospect, an EUS-guided biopsy might have established the diagnosis preoperatively. However, given the high suspicion and the need for definitive treatment, proceeding directly to surgical resection was a reasonable approach in our patient. Preoperative tissue diagnosis, while ideal, is not always mandatory for resectable esophageal masses, especially when the differential includes a potentially curable tumor and the risks of surgery are justified.

Imaging characteristics: Our patient's CT findings were fairly typical for a leiomyosarcoma in several respects. Leiomyosarcomas of the esophagus are often large, lobulated masses with internal necrosis or cystic areas (due to outgrowing their blood supply). On CT they tend to appear as heterogeneous masses that may have both intraluminal and extraluminal components. In our case, the tumor was predominantly extraluminal (paraesophageal) with a necrotic center, and it caused extrinsic compression of the esophagus rather than an intraluminal obstruction. No gross luminal tumor was seen on endoscopy, concordant with the imaging. This pattern aligns with the "infiltrative/intramural" type leiomyosarcoma (as opposed to a polypoid intraluminal type). Rainer and Braus have historically classified esophageal leiomyosarcomas into two growth patterns: polypoid (60%) which protrude into the lumen, and infiltrative (40%) which grow within the wall and outward. Our case falls into the infiltrative category. This distinction is prognostically relevant, as discussed below. Importantly, the CT also demonstrated indirect signs of RLN palsy (atrophy of left laryngeal muscles), an unusual but useful radiologic clue correlating the patient's symptom with the mediastinal mass. This multidisciplinary correlation of radiologic and clinical findings was crucial in steering the diagnosis toward an esophageal leiomyosarcoma even before surgery.

Pathology and differential diagnosis: Histologically, esophageal leiomyosarcoma must be distinguished from other spindle cell tumors of the esophagus. The differential diagnosis includes gastrointestinal stromal tumor (GIST), leiomyoma (the benign counterpart), and rarely schwannomas or other sarcomas. Immunohistochemistry is essential for this distinction. Leiomyosarcomas characteristically express smooth muscle markers such as α -smooth muscle actin, desmin, h-caldesmon, and calponin. They do not express KIT (CD117) or DOG1, which are typically positive in GISTs. In our patient, the immunoprofile (SMA and desmin positive, KIT/DOG1 negative) was consistent with leiomyosarcoma. Esophageal GISTs are extremely uncommon, but they tend to occur in the distal esophagus and often have mutations in KIT or PDGFRA; thus, distinguishing these entities is important as it impacts therapy (GISTs are treated with imatinib-targeted therapy, whereas leiomyosarcomas are not). The benign esophageal leiomyoma is far more common than leiomyosarcoma in the esophagus, but leiomyomas are usually smaller, symptomatic only if large, and of course lack the atypia and mitoses of leiomyosarcoma. In our case, the tumor's high mitotic count and invasive behavior confirmed its malignancy. Another rare consideration is a dedifferentiated carcinoma (carcinosarcoma or spindle cell carcinoma) of the esophagus, which can have a spindle component. However, those usually have at least focal epithelial components (and often present more like typical carcinoma). No epithelial elements were seen in this case, arguing against a spindle cell carcinoma. We also note that a synchronous esophageal squamous carcinoma coexisting with leiomyosarcoma has been reported in the literature (so-called "kissing tumors"), but this was not observed here.

Treatment considerations: Surgical resection is the mainstay of treatment for esophageal leiomyosarcoma and offers the only potential cure. Given the rarity of the disease, there are no

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randomized trials, but case series consistently emphasize complete surgical excision as the optimal strategy. In our patient, a transthoracic esophagectomy achieved a complete resection. An important surgical consideration is that some leiomyosarcomas, especially polypoid types confined to the esophagus, might be amenable to more limited resection (e.g., enucleation or segmental resection) if diagnosed preoperatively as benign or low-grade lesions. In fact, historical cases report simple local excisions for well-localized tumors with good outcomes. However, in most modern reports, an esophagectomy (with reconstruction) is performed, especially for larger or invasive tumors, to ensure clear margins. In the Mayo Clinic series of 17 patients, a variety of surgical approaches were employed (including enucleation in a few cases and esophagectomy in others), and achieving clear margins was associated with improved survival.

The role of adjuvant therapy (chemotherapy or radiotherapy) in esophageal leiomyosarcoma is not well established and remains controversial. Unlike soft tissue sarcomas of the extremities, where adjuvant radiation is often used to improve local control, the use of radiation in the esophagus is complicated by the proximity of radiosensitive organs (spinal cord, lungs, etc.) and the risk of severe esophagitis. Chemotherapy regimens for leiomyosarcoma (usually protocols for soft tissue sarcomas using agents like doxorubicin and ifosfamide) have been tried in metastatic or unresectable cases, but their efficacy is limited. Small case reports and series have documented instances of using radiotherapy for residual or inoperable esophageal LMS, with occasional disease control, but there is no clear survival benefit in the adjuvant setting. In our case, since a complete resection was achieved and the patient had no evidence of spread, we did not pursue adjuvant therapy. This decision is in line with literature suggesting that adjuvant chemo/radiotherapy does not significantly improve outcomes in completely resected leiomyosarcoma of the esophagus. The patient's excellent disease-free survival at 3 years post-surgery supports the adequacy of surgery alone in this scenario.

Prognosis: Overall, esophageal leiomyosarcoma has a better prognosis than the more common esophageal squamous cell carcinoma. Several factors influence outcomes: tumor size, grade, depth of invasion, and growth pattern (polypoid vs infiltrative) have been identified as prognostic indicators. Polypoid, intraluminal tumors that are well-differentiated tend to be detected earlier (due to dysphagia) and have a more favorable outcome. Infiltrative tumors (like in our patient) or those located in the cervical esophagus are associated with a higher risk of local extension and positive margins. In one surgical series, 5-year survival was 74% for patients with intramural/polypoid growth versus only 14% for those with infiltrative growth. Likewise, complete resection (R0) is crucial: patients who undergo curative resection have significantly better survival than those with incomplete resection or metastases. Reported 5-year survival rates for resected esophageal leiomyosarcoma range from about 30% up to 60% in various series, which, while lower than the 74% cited for polypoid tumors, are still notably higher than 5-year survival in esophageal squamous carcinoma. Our patient's course – disease-free at three years – is encouraging, although continued surveillance is needed. Approximately one-third of esophageal leiomyosarcomas have already metastasized at the time of diagnosis, most commonly to the liver or lungs. Fortunately, our patient had no metastasis on workup. There is also a risk of late recurrence; cases of tumor recurrence many years after resection have been noted, so long-term follow-up is recommended. Given the high-grade nature of our patient's tumor, we plan to monitor him for at least 5–10 years postoperatively.

Literature review: Reviewing the literature, we found only a handful of cases that shared features with this one. Eroglu et al. (2015) reported a 48-year-old woman with a large esophageal leiomyosarcoma who was treated definitively with radiotherapy, remaining

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disease-free at 2 years – an unusual non-surgical management. Koga et al. (1995) described a rapidly growing esophageal leiomyosarcoma with a poor outcome, suggesting that coexistence with a carcinoma component can accelerate sarcoma progression. Rocco et al. (1998) presented the largest single-center experience, reinforcing that complete surgical excision yields the best outcomes. Notably, a recent review by Haidrus and Mittal (2025) echoed many of these points: they emphasized the importance of considering leiomyosarcoma when endoscopy is unrevealing but imaging suggests an esophageal mass, and they highlighted that long-term survival is achievable in these rare tumors, often outpacing that of typical esophageal cancers. Our case contributes to the literature by illustrating the scenario of an infiltrative upper esophageal leiomyosarcoma that presented with RLN palsy and great vessel involvement, yet was successfully resected and resulted in an excellent patient outcome. It underscores the need for multidisciplinary evaluation – involving otolaryngologists, gastroenterologists, radiologists, and surgeons – in diagnosing and treating unusual presentations of esophageal tumors.

Learning Points

- Unusual presentations of esophageal tumors: Esophageal leiomyosarcoma, though rare, should be considered in patients with unexplained vocal cord paralysis or mediastinal masses. Hoarseness due to left recurrent laryngeal nerve palsy can be a presenting sign of an upper esophageal or mediastinal tumor, even in the absence of the typical symptom of dysphagia.
- Diagnostic approach to submucosal masses: A normal endoscopic mucosal biopsy does not rule out an esophageal tumor if clinical suspicion remains high. Submucosal lesions can be missed by superficial biopsies. In such cases, advanced diagnostic tools like endoscopic ultrasound-guided biopsy should be utilized, or empiric surgical exploration may be warranted for definitive diagnosis and treatment.
- Management and prognosis: Complete surgical resection of esophageal leiomyosarcoma offers the best chance for cure and long-term survival. Encasement of adjacent structures (e.g. great vessels or nerves) on imaging does not always preclude resection, as these tumors often dissect planes without widespread invasion. Adjuvant therapy has an uncertain benefit in this disease, so surgery-first approach is standard. Overall prognosis for localized leiomyosarcoma of the esophagus is more favorable than that of esophageal carcinoma, but factors such as infiltrative growth pattern, high grade, or incomplete resection portend a higher risk of recurrence, necessitating careful long-term follow-up.

Patient Perspective

No patient perspective is available. (The patient declined to provide a personal statement, but he has expressed gratitude for the care he received and is pleased with his recovery and current quality of life.)

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