Case report – Thoracic general

Benign intramural schwannoma of the oesophagus: a diagnostic pitfall

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Abstract

A 33-year-old man underwent resection of a benign intramural schwannoma of the oesophagus. Similar clinicopathological and immunohistological features prompted a review of an earlier case initially diagnosed as a S-100 negative leiomyosarcoma. This stained positive using a new S-100 immunohistochemical antibody (Dako, Trappes, France). The frequency of these extremely rare tumours may be higher than initially thought. Pitfalls for the surgeon are highlighted and attention is brought to a recent consensus.

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1. Case report

A 33-year-old previously healthy man presented with limiting dyspnoea and wheeze, two-years of cough and dysphagia, 16 kg weight loss in 4 months and regurgitating undigested food at every meal. Inspiratory stridor was noted. There were no clinical signs of neurofibromatosis. Barium swallow suggested a large intramural mass displacing and compressing the lumen of the oesophagus. Computed tomography (CT) located a concentric soft tissue lesion surrounding the oesophagus, starting at the level of the suprasternal notch, running between the vertebral bodies and the trachea to the carina (Fig. 1). Bronchoscopy confirmed severe narrowing of the tracheal lumen throughout its intrathoracic course. Oesophagoscopy revealed an intraluminal mass with a sharp transition to normal mucosa raising the possibility of an intraluminal polyp\textsuperscript{[2]}. The biopsies showed only that the overlying mucosa was necrotic.

At right thoracotomy an 8.2 $\times$ 5.0 $\times$ 3.6-cm multilobulated firm, rubbery, pale white tumour was found related to the muscular wall of the oesophagus. The tumour was scalloped and contoured by the vertebral bodies. It displaced the trachea anteriorly and was adherent to its posterior wall but there was no apparent invasion. The mass was enucleated within the wall of the oesophagus and was found to have eroded from the mucosa into the oesophageal lumen. Frozen section biopsy showed presence of a spindle cell tumour with pleomorphism but no evidence of mitoses, suggesting a low-grade leiomyoma or leiomyosarcoma. The mucosal disc excised with the tumour showed erosion of tumour immediately beneath the epithelium but no clear evidence of invasion. The mass had expanded the oesophageal wall allowing primary closure of the mucosal and muscular layers after resection and the oesophagus was conserved. Benign schwannoma was diagnosed only after the paraffin and immunological staining was examined.

The clinical similarity led to a review of an earlier case in which mucosal involvement by a spindle cell tumour had led to oesophagectomy. Initially classified as a low-grade leiomyosarcoma postoperatively, the diagnosis of benign schwannoma was subsequently confirmed with microscopy, immunohistochemical studies and electron microscopy, the features of which are described below. Both patients are alive and well at 3 and 15 years later with no evidence of recurrence.

2. Discussion

Neural tumours of gastrointestinal tract origin can be confused with smooth muscle tumours (leiomyomas or leiomyosarcomas) in the oesophagus and their clinical, radiological and histopathological appearances may lead one to suspect malignancy\textsuperscript{[1,3]}. Daimaru et al. suggested that misdiagnoses occurred before immunohistochemical analysis was developed \textsuperscript{[4]}. Our second case, in retrospect essentially a misdiagnosis, confirms the inaccuracy of
earlier tests for immunostaining S-100 proteins. This problem is exacerbated by the elusiveness of diagnostic pre-operative tissue biopsies, as experienced in this case report, often only revealing overlying necrotic mucosa.

Benign intramural oesophageal schwannomas are remote from the vagus nerve, have a spindle cell structure with no epithelioid features and a prominent lymphoid cuffing surrounding the tumour [6]. Electron microscopy shows
ultrastructural features of well-developed external basal lamina and intra cellular needle shaped PAS-positive crystalloids [3–6]. Recently, using comparative genomic hybridization (CGH), Sarlomo-Rikala et al. showed that schwannomas are genetically different to gastrointestinal stromal tumours (GISTs), leiomyomas and leiomyosarcomas. 77% of GISTs studied (n = 13) showed DNA copy losses in 14q and 62% additional or other losses in chromosomes 22, 15 and 1p. Only one case of schwannoma and 3 cases of leiomyomas showed DNA changes using CGH [7]. Miettinen et al. showed that Schwannomas are negative for CD34 and CD117 (c-kit protein), whereas GISTs and gastrointestinal autonomic nerve tumours which no longer warrant designation as a separate entity from GIST [1] are positive for both [8].

The authors would like to draw attention to the recent National Institutes of Health consensus review [1] in which the confusion and controversy surrounding diagnostic pitfalls, such as this case, has been addressed comprehensively. The immunohistochemical schema for the differential diagnosis of spindle cell tumours of the GI tract (Table 1) agreed in this consensus has clarified the diagnostic process. Retrospective review of previous cases using the schema may reveal higher incidences of benign intramural schwannomas of the oesophagus or gastrointestinal tract, previously misdiagnosed as leiomyomas or leiomyosarcomas, and differentiate them from potentially malignant GISTs [1].

The surgeon should be aware that diagnostic pre-operative tissue biopsies are elusive, and that the lengthy immunohistochemical examinations required to differentiate this complex diagnosis are unavailable at the time of surgery. This focuses the surgeon’s decision, regarding surgical options, on the appearance of the tumour at the time of surgery. The value of frozen section is at present limited in the information it can supply to the surgeon with regards to this diagnostic pitfall. The option of conservative enucleation and thus avoiding the morbidity of oesophageal resection is preferable provided: the tumour is well encapsulated, a clear margin is achievable, and that there is sufficient redundant mucosa to close the defect. Without an achievable clear margin, oesophageal resection is indicated as with the earlier case in this series. Long-term prognosis after conservative resection of benign oesophageal Schwannoma is unknown.

Table 1

<table>
<thead>
<tr>
<th></th>
<th>KIT (CD117)</th>
<th>CD34</th>
<th>SMA</th>
<th>Desmin</th>
<th>S-100</th>
</tr>
</thead>
<tbody>
<tr>
<td>GIST</td>
<td>+</td>
<td>+ (60–70%)</td>
<td>+ (30–40%)</td>
<td>Very rare</td>
<td>5% +</td>
</tr>
<tr>
<td>Smooth muscle tumor</td>
<td>−</td>
<td>+ (10–15%)</td>
<td>+</td>
<td>+</td>
<td>Rare</td>
</tr>
<tr>
<td>Schwannoma</td>
<td>−</td>
<td>+ (usually Antoni B)</td>
<td>−</td>
<td>−</td>
<td>+</td>
</tr>
<tr>
<td>Fibromatosis</td>
<td>Disputed</td>
<td>Rare</td>
<td>+</td>
<td>Rare cells</td>
<td>−</td>
</tr>
</tbody>
</table>

SMA, smooth muscle actin.

a Most, but not all authors report that fibromatoses are negative for KIT.

References


Appendix A. ICVTS on-line discussion

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Message: This article highlights an important aspect of surgical practice, and I congratulate the authors for raising this issue. Although, histopathological diagnoses are not within the control of the surgeon, it dictates his practice to a large extent, and consequently surgical outcomes. Quite rightly, surgeons are getting more involved in reaching the diagnosis in such difficult cases. Our experience with an oesophageal melanotic schwannoma (Thoracic and Cardiovascular Surgeon 2002;50:103-104) lends credence to the elusiveness of the diagnosis of gastrointestinal schwannomas.