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CASE REPORT

Achalasia leading to diagnosis of adenocarcinoma of the oesophagus

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SUMMARY

A 50-year-old male with a 7 month history of progressive dysphagia to solids then subsequently to liquids. He underwent a diagnostic gastroscopy which was normal. A further barium swallow suggested achalasia. He was referred to a tertiary centre, where he underwent pH and manometry studies which confirmed a diagnosis of achalasia. He was referred for a laparoscopic cardiomyotomy, and at surgery there was a suspected tumour at the gastro-oesophageal junction. A follow-up endoscopy with biopsies was normal. Following this, a positron emission tomography scan showed T3 distal oesophageal cancer with no nodal involvement or distal metastasis. An attempt at oesophagectomy was performed, but at operation there was locally advanced carcinoma infiltrating the coeliac axis. He is currently undergoing palliative chemotherapy.

BACKGROUND

Oesophageal cancer is the 13th most common cancer in the UK (2011), accounting for 3% of all new cases. In males, it is the eighth most common cancer, while it is the 14th most common in females.¹⁻⁴

In England, the most common oesophageal cancers are adenocarcinomas which account for more than half (55%).¹ The second most common oesophageal cancers in England are squamous cell carcinomas, which account for more than a quarter (28%).¹

The risk factors for the development of squamous cell carcinoma of the oesophagus have been identified to include tobacco, alcohol and diet⁵; however, the risk factors associated with oesophageal adenocarcinoma are less clear.⁵

Oesophageal symptoms include dysphagia, pain, vomiting and weight loss. When symptoms are present, often the first step in aiding the diagnosis is to undergo gastroscopy. The sensitivity of gastroscopy for oesophageal cancer has been reported at 94.5%.⁶ Other less used methods for diagnosis are barium swallow and CT-guided biopsies; however, endoscopic evaluation remains the gold standard.

Despite these sensitive investigations, the disease is often at an advanced stage when symptoms are present, meaning oesophageal cancer is rarely curable. Of all those diagnosed with oesophageal cancer, 42% will live for at least 1 year after they are diagnosed, 15% will live for at least 5 years and 12% will live for at least 10 years. The outcome of oesophageal cancer depends on how advanced it is

at diagnosis.⁷ Therefore, making an early diagnosis is paramount to aid survival chances.

There are many conditions that may mimic some of the symptoms of oesophageal cancer. One of these conditions is called achalasia, defined as a failure of the smooth muscle fibres to relax in the lower oesophagus. To our knowledge, achalasia as a diagnosis is not associated with a diagnosis of oesophageal cancer. It has been reported, however, that over time achalasia is a risk factor for the development of oesophageal cancer, with the risk reported at 0.4% to 9.2%.⁶

There are case reports noting a rare condition called pseudoachalasia. This is defined as signs and symptoms consistent with primary achalasia but which is due instead to invasion of the distal oesophagus or its neural plexus by malignant tumours. Pseudoachalasia has been mostly associated with gastric adenocarcinomas or squamous cell carcinomas of the oesophagus but rarely as adenocarcinomas of the oesophagus.⁸

To our knowledge, there is one case report highlighting pseudoachalasia as an only finding in an adenocarcinoma and this was in a juvenile with polyposis syndrome.⁹ We believe this is a rare finding; therefore, we present a case report where achalasia has been the only symptom in adenocarcinoma of the oesophagus.

CASE PRESENTATION

A 50-year-old British man presented with a 7 month history of progressive dysphagia to both solids and liquids. He initially found it difficult to swallow meats and breads which progressed over 3 months to liquids. In association with this, he had lost 20 kilograms in weight. He also started to notice some epigastric discomfort.

He had no medical history of note. At time of presentation, he was not on any regular medications with no reported drug allergies. He worked as a lorry driver, smoked 10–20 cigarettes a day with a 20 pack year history. He consumed 1–2 units of alcohol a week. He lived with his wife and was fully independent. There was no family history of any significance.

INVESTIGATIONS

He was originally referred as a direct access to endoscopy case via the GP on 13 September 2012. This gastroscopy was normal to the second part of the duodenum. Following a review a month later in the gastroenterology clinic, he was referred for a



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Unusual presentation of more common disease/injury

barium swallow. This showed a distal oesophagus that was seen to taper just before the gastro-oesophageal junction which was consistent with achalasia. While awaiting surgical treatment, he was referred for confirmation of achalasia by manometry studies at a tertiary centre. This demonstrated a high-pressure, non-relaxing lower oesophageal sphincter and a complete absence of peristalsis in the oesophageal body. The findings were consistent with achalasia type 2. Following a suspicious laparoscopic cardiomyotomy, the patient was referred for a repeat gastroscopy to try and get some tissue for histology. The biopsy samples from the distal gastro-oesophageal junction showed no malignant tissue. He then underwent a CT scan of the chest and abdomen which showed a dilated fluid-filled oesophagus at the level of the gastro-oesophageal junction. The CT showed no other visceral abnormalities in the chest or abdomen.

He then underwent a positron emission tomography (PET) scan which highlighted a T3 distal oesophageal tumour but with no nodal involvement. He was planned for an oesophagectomy with insertion of a feeding jejunostomy. Unfortunately, this could not be completed due to findings of locally advanced disease. At the time, biopsies were taken from peripancreatic lymph nodes which confirmed metastatic adenocarcinoma.

He was tested for HER2 receptor and was positive for this.

DIFFERENTIAL DIAGNOSIS

Dysphagia as a symptom can be caused by a variety of conditions both intraluminal, luminal and extraluminal. Intraluminal causes include masses and food boluses. Luminal include strictures both benign and malignant, oesophagitis, infection and neurological abnormalities including achalasia. In someone who can tolerate it, endoscopy is usually the first step in trying to differentiate the different causes. In this case, the endoscopy was normal. To try and rule out some of the other causes such as achalasia, a barium swallow can offer a simple and minimally invasive next step. With a normal endoscopy and normal biopsies, most intraluminal and luminal causes can be excluded. Barium swallow can highlight pouches and can be suggestive of achalasia with the typical 'bird beak' appearance. In this case, the barium swallow was suggestive of achalasia. The finding of achalasia is usually confirmed via manometry studies. We referred externally for pH and manometry studies. Achalasia was confirmed when these tests were done.

From this point with ongoing symptoms, we planned to treat the achalasia with a Heller's myotomy procedure. It was only at this operation that a cancer was found and therefore was referred for oesophagectomy.

TREATMENT

The patient was referred for a laparoscopic cardiomyotomy. At operation, there was a suspicious mass at the gastro-oesophageal junction. Following a CT of the chest and abdomen which showed no visceral abnormalities to suggest cancer, he underwent laparoscopic staging including sending fluid for cytology. At this stage, there was no evidence of disseminated disease. He then underwent a further endoscopy for histological confirmation. Following a repeated endoscopy and PET scan, he was referred for an attempt at oesophagectomy. Unfortunately, at time of the operation, locally advanced carcinoma which had infiltrated the coeliac axis was found. Histology taken confirmed metastatic adenocarcinoma. At the time of this admission, he had an oesophageal stent inserted and dilatation to help his dysphagia. The cancer expressed HER2. He was then started on palliative chemotherapy. He responded well to this and after

a multidisciplinary team discussion, he was started on cisplatin and capecitabine. He tolerated this quite well. The most recent conversation with the patient reveals he remains stable on chemotherapy.

OUTCOME AND FOLLOW-UP

The patient remains on chemotherapy. He remains in regular contact with the oncologist and on last discussion remains active and is well.

DISCUSSION

To our knowledge, new onset diagnosis of achalasia has not been reported as a symptom of adenocarcinoma of the oesophagus. It is, however, known that long-standing achalasia can lead to squamous cell carcinoma of oesophagus, but there is no mention in the literature of adenocarcinoma. The risk for development of oesophageal cancer is 0.4% to 9.2% in achalasia.¹⁰ Adenocarcinoma of oesophagus is only reported in achalasia patients who have undergone pneumatic dilation, due to gastro-oesophageal reflux leading to Barrett's metaplasia.¹¹

Learning points

- ▶ Recurrent dysphagia not responding to treatment should be investigated thoroughly.
- ▶ Achalasia can mask an underlying distal oesophageal cancer.
- ▶ Long-standing achalasia can lead to squamous carcinoma, whereas the development of adenocarcinoma has not been previously reported.

Patient's perspective

It all started by feeling blocked up when eating and being sick. There was a considerable delay both at my GP and the hospital and eventually I had an operation which then showed instead of having achalasia I had cancer. I went in for major surgery which was unsuccessful. I have had chemotherapy and still have Herceptin every 3 weeks together with various scans and a recent blood transfusion. I receive excellent treatment now, but there have been many frustrations. I will never know if the delays early on in my diagnosis contributed to where I am now. I call myself a ticking time bomb and continue to try and live my life as normally as possible under the circumstances.

Contributors JS did the initial literature search around the topic, contacted the patient for consent and discussion. He gathered all the data and analysed the data. He wrote the manuscript and edited it. AL helped refine the manuscript. MC was the primary physician of the patient and helped with the planning of the manuscript including conception and design. He also helped with refinement of the manuscript.

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