Diffuse oesophageal leiomyomatosis

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Giant oesophageal leiomyomas are rare benign neoplasms accounting for 0.4% of all oesophageal tumours. Diffuse leiomyomatosis is an even rarer entity among oesophageal diseases, with only approximately 40 cases reported in the literature.[1] In contrast to a leiomyoma, which typically comprises a discrete tumour nodule, leiomyomatosis is characterized histopathologically by diffuse hypertrophy of the muscular layer.

A 31-year-old woman presented with a 1-year history of vague chest discomfort. She was otherwise asymptomatic. In her past surgical history, she had bilateral congenital cataract repair when she was 20 years old. Her initial presentation prompted her doctor to request a chest radiograph (CXR). Unexpectedly, her CXR showed a large mediastinal mass measuring 9 cm (Fig. 1). A computed tomography scan revealed a huge mass in the distal oesophagus extending just below the diaphragm. The mass was oval-shaped and could be seen posterior to the cardiac border. Initial oesophagogastroduodenoscopy and biopsies suggested a benign leiomyoma.

Figure 1.

Chest radiograph demonstrating a large mediastinal mass measuring 9 cm.

She was referred to the National Centre for Upper Gastrointestinal Surgery at St. James's Hospital for further assessment. Her physical examination was normal. A positron emission tomography-computed tomography scan demonstrated a large oesophageal tumour measuring 10×7 cm in maximum cross-sectional dimensions, showing increased metabolic activity with a maximum standardized uptake value (SUVmax) of 7.7. The tumour extended from the lower oesophagus and involved the oesophago-gastric junction, but there was no evidence of disease elsewhere. An endoscopic ultrasound showed an oesophageal mass beginning at 21 cm, at the level of the aortic arch, extending into the posterior mediastinum, 7 cm in size, being hypoechoic and heterogeneous on ultrasound. The lesion was mostly solid in nature and was emanating from the wall of the oesophagus. Biopsies showed benign smooth muscle.

This tumour clinically and radiologically resembled a gastrointestinal stromal tumour, but biopsies suggested a leiomyoma. Because of the patient's ongoing chest discomfort, and the potential for malignancy, it was decided that surgery was the most appropriate intervention. At surgery, a highly vascular mass was evident from the mid-oesophagus extending to the hiatus, where it reached maximal dimensions (Fig. 2). A left oblique neck incision allowed for mobilization of the cervical oesophagus, transection and an end-to-end oesophago-gastric anastomosis. She was admitted to the high dependency unit post-operatively, commenced enteral feeding via a jejunostomy on day 1 post-op and had an uneventful recovery. Histopathological examination of the specimen revealed multiple confluent nodules of tumour within the muscularis propria (Fig. 3). Diffuse thickening of the muscularis mucosae was also seen. The lesion was positive for smooth muscle actin and desmin but was negative for CD117 (c-Kit) and DOG-1. This was concurrent with a diagnosis of diffuse oesophageal leiomyomatosis.

Figure 2.

Operative photograph of the highly vascular oesophageal mass.

Figure 3.

Macroscopic examination of the resection specimen showed multiple nodules in the oesophageal wall, the largest being 15 cm in size (image on left). The cut surface of the nodules showed a pale whorled appearance (image on right).

Oesophageal leiomyomatosis may occur sporadically or on a hereditary basis with autosomal dominant inheritance.[1, 2] It may also have an association with other gastrointestinal leiomyomata (particularly small intestine and rectal) along with widespread visceral leiomyomatosis, tracheobronchial lesions and genital lesions in women, including clitoral hypertrophy and vulval leiomyomatosis, which constitutes the oesophagovulvar syndrome.[3, 4]

Oesophageal leiomyomatosis typically presents in children and young adults and is slightly more common in men.[2] Dysphagia and pain, as in our patient, are the most common presenting symptoms followed by weight loss, nausea and vomiting.[1] Less frequent symptoms include recurrent pneumonia, respiratory symptoms and dyspnoea. Up to 50% of patients are asymptomatic at the time of diagnosis and are discovered incidentally, typically on a CXR, for another indication.[1, 2] Partial or subtotal oesophageal resection is recommended, as in this case, as these tumours are often very large.[3-5] Standardized lymphadenectomy is not necessary given that malignant transformation does not occur.[5]

Alport syndrome is a progressive, heterogeneous nephropathy characterized by the association of progressive haematuric nephritis, sensorineural deafness and variable ocular abnormalities (bilateral congenital cataracts, anterior lenticonus and macular flecks).[6] It has an incidence of approximately 1 in 5000 individuals and is more common in men. Diffuse leiomyomatosis occurs in 5% of cases of Alport syndrome; however, other studies report that Alport syndrome can be present in greater than 72% of children with oesophageal leiomyomatosis.[7] Our patient did not have any known personal or family history of Alport syndrome. Her history is significant, however, for bilateral congenital cataracts, which have been independently associated with oesophageal leiomyomatosis in some cases and may share some of the same genetic associations with Alport syndrome; however, in our patient, the renal function and urinary microscopy was normal.[2, 4, 6, 8] Whether isolated or in association, diffuse oesophageal leiomyomatosis remains a benign disorder.[2] There are no associations with neoplastic transformation in the literature and radical surgical treatment is typically curative.[2]

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